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Section One of Two Parts

Vol. 3 March, 1956 No. 1

FERTILITY CONTROL IN THE LIGHT OF SOME CATHOLIC STATEMENTS
William J. Gibbons, S.J.

TREATMENT IN HEREDITARY DISEASES
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THE PROSPECTS FOR GENETIC IMPROVEMENT
L. C. Dunn

HEREDITARY SUSCEPTIBILITY IN RHEUMATIC FEVER
May G. Wilson, M.D.

American Eugenics Society, Inc.

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POPULATION STUDIES

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Edited by
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Fabian Bachrach

DR. HARRY L. SHAPIRO
Newly Elected President of the American
Eugenics Society

Vol. 3, No. 1, March, 1956 *Eugenics Quarterly*

EDITORIAL COMMENT

New Officers of A.E.S. Elected

Dr. Harry L. Shapiro, Chairman of the Department of Anthropology at the American Museum of Natural History, has been elected President of the American Eugenics Society to serve from 1956 through 1958. A director of the Eugenics Society for many years, he has just completed a three-year term as its Vice-President.

Dr. Shapiro is also Professor of Anthropology at Columbia University and Research Associate in Anthropology at the Bishop Museum, Honolulu. A past president of the American Anthropological Association and the American Ethnological Association, he is a member of the National Academy of Sciences. He was chairman of the Division of Anthropology and Psychology of the National Research Council from 1932 to 1933. His chief fields of research are physical anthropology, human biology, race mixture, and population. Field work was carried on in Polynesia, Puerto Rica, the Orient, the eastern Pacific, Alaska, and the southwestern United States.

In addition to contributions to many scientific journals, Dr. Shapiro has published two books: "The Heritage of the Bounty" (1936) and "Migration and Environment (1939).

Alan F. Guttmacher, M.D. head of the Department of Obstetrics and Gynecology at The Mount Sinai Hospital in New York City, is the newly elected Vice-President of the Eugenics Society. He is also clinical professor of Obstetrics and Gynecology at Columbia Medical School. He was formerly chief of Obstetrics at The Sinai Hospital of Baltimore.

A diplomate of the Board in Obstetrics and Gynecology, he is a fellow of the Academy of Obstetrics and Gynecology and the American Society for the Study of Sterility. He is an associate editor of *Fertility and Sterility* and Vice-President of the Planned Parenthood Federation of America from whom he received the Lasker Award in 1947.

Dr. Guttmacher has published extensively on infertility, contraception, induction of labor, and the history of medicine. He is the author of the pocketbook, "Having a Baby."

Chauncey Belknap, lawyer, was re-elected Treasurer. Mr. Belknap is a member of the firm of Patterson, Belknap, and Webb in New York City and has been a Director of the Eugenics Society for many years.

Frederick Osborn, demographer of New York City, was re-elected Secretary.

Directors of the Society will be elected at the Annual Meeting. Suggestions for candidates should be sent to the Chairman of the Nominating Committee, American Eugenics Society, 230 Park Avenue, New York 17, N. Y.

Editor's Note:

Comment by Robert C. Cook and Frederick Osborn on The Eugenic Hypothesis has been published in EUGENICS QUARTERLY, *September 1955, pp 129-132 and December, 1955, pp. 194-197.*

The following letters continue this discussion. Copies of the original presentation of The Eugenic Hypothesis by Frederick Osborn are available upon request to the Editor. Readers are invited to contribute their comments.

To the Editor:

I have read with interest the exchange of comment concerning the validity of the Eugenic Hypothesis (Cook, 1955 a, b; Osborn, 1955). Although the main objections of Robert C. Cook are now admitted to have been based on a misunderstanding, the same misunderstanding and similar objections may still linger in the mind of some skeptics. In any event, the genetic basis of the Eugenic Hypothesis seems to deserve closer scrutiny than it has received up to now, and I think this scrutiny will reveal no points of conflict with present genetic theory.

The Eugenic Hypothesis is expressed by its author as follows (Osborn, p. 240): "If the environment from childhood includes those influences which make for full development of the desire for children, then those individuals most susceptible to the stimulus of the environment will tend to have more children than will their less responsive fellows. . . . The response would be voluntary, a selection drawn out by the environment itself, so that in so far as children tend to resemble their parents for either hereditary or environmental reasons, there would be in each generation a proportionate increase in the number of people best fitted to the environment."

Cook objected, first, to "the assumption that 'group inheritance'—a transmission *en bloc*—of a whole constellation of desirable characteristics is a biological possibility." As Cook understood it, the Eugenic Hypothesis required *inheritance* of the desire for children and, further, linkage of this with all valuable genetic qualities. Quite the contrary! The Eugenic Hypothesis depends upon the possibility of *determining* the desire for children through socio-cultural influences. As I see it, there are only two other major assumptions implicit in the Eugenic Hypothesis. One is that responsiveness to our socio-cultural environment is partly determined by genetic variations within the species. The other is that this responsiveness itself is the trait of primary interest for positive eugenics. None of these assumptions appears to be seriously disputed.

According to the Eugenic Hypothesis, desire for children and responsiveness to the environment will be linked not genetically, but by the deliberate creation of a *Eugenic Environment.* This environment would be designed to stimulate the desire for children in persons most responsive to their total environment, without influencing those whose interests were narrower or whose physical and mental handicaps made child-rearing too burdensome. Of course, the Eugenic Environment can achieve its purpose only if, at the same time, adequate contraception makes fertility dependent upon desire for children (*vide infra*).

A second objection was phrased by Cook as follows: "There is a further hard-to-define Lamarckian flavor to the Eugenic Hypothesis that seems to run counter to biological realities." Before we argue about this "hard-to-define" flavor, let us make some effort to define it! Lamarckianism is the doctrine that acquired characters are biologically heritable. There are two points at which such a doctrine is suggested by the Eugenic Hypothesis. First, Osborn says that *even in so far as children resemble their parents for environmental reasons, a eugenic environment would multiply the people best fitted to it.* It is not truly Lamarckian to believe in social inheritance of acquired behavior patterns. Competition and selection among cultures can operate independently of and even in opposition to

biological natural selection. Families that have, for purely environmental reasons, developed a superior pattern of social adaptation are likely to transmit this pattern by precept and example from one generation to the next. If such families were to multiply more than others in a given environment, well-adapted people would eventually predominate even if genetic progress remained at a standstill. This is surely a desirable result in itself, and probably as deserving of our efforts as genetic progress. The eugenics movement does, in fact, seem to be broadening its goals to include cultural improvement of the population in so far as this does not conflict with genetic improvement.

The second point in the Eugenic Hypothesis which resembles Lamarckianism is the implication that *socially desirable parents are the most likely to have children with a genetic capacity for becoming socially desirable.* This implication, however, follows directly from the concepts which geneticists have used to combat Lamarckianism (Schmalhausen, 1949; Waddington, 1953). People who acquire socially desirable behavior patterns (even including the phenocopies mentioned in Cook's second communication) evidently have the genetic capacity to do so, while among those who fail to acquire desirable patterns will be found *all* those who lack the requisite genetic capacity. If a Eugenic Environment can be devised that will increase fertility among people with socially desirable traits relative to fertility in those without them, this environment will *pari passu* select desirable genetic capacities and increase the proportion of the population genetically capable of acquiring the most desirable behavior patterns.

Finally, Cook finds support for his argument in observation. "Even a casual exploration of the world about us should strengthen our doubts that such a force is operating." This is where we came in! Eugenists all became eugenists, I suspect, because they feared that overall selection is now operating in the wrong direction. Yet in his second communication Cook developed this point further: ". . . the question may be raised as to the extent to which a selection program based on such vague criteria is really going to be effective in reversing the counter-selective trends which the very recent pattern of mortality now obtaining in 'advanced' nations appears to make inevitable." Calling for a realistic appraisal of any positive fertility differentials currently associated with desirable qualities, he suggests that their eugenic influence cannot be very effective.

There are in fact four propositions here which must be carefully separated. First, probably no modern eugenist would maintain that the eugenic differentials now indicated within certain very limited groups are compensating the much more prevalent dysgenic differentials.

Second, the Eugenic Hypothesis does not imply that genetic progress would be insured even by extending to all classes these within-group differentials that seem to be associated with successful family planning. Universal "freedom of choice for parenthood" would probably remedy some of the problems that stir eugenists, but it might aggravate others.

Third, it is hardly reasonable to debate whether Osborn's Eugenic Environment would, in fact, be eugenic. This environment has yet to be created, but, by definition, no environment will meet the requirements if it does not reverse or offset present dysgenic trends. It is legitimate to ask whether man will ever be able to create for himself an environment that is eugenic in Osborn's sense. This question is recognized and explored in *Preface to Eugenics.*

Fourth, if the Eugenic Hypothesis is ever proved, there will probably still be no precise reversal of present counter-selective trends. It is not necessary that "birth selection" in the Eugenic Environment should fill the exact role that "death selection" did in the environment of our ancestors. Overall evolutionary progress is often

achieved in spite of, or even made possible by, genetic retrogression in certain spheres. It does not matter too much if some hereditary diseases multiply, provided that hereditary factors for intelligence and social usefulness multiply faster. These are the traits which would be most favored by natural selection in Osborn's Eugenic Environment.

GORDON ALLEN, M.D.
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New York, N. Y.

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WADDINGTON, C. H. 1953. (December). Experiments in acquired characteristics. *Scientific American*, pp. 92-99.

To the Editor:

My comment on Dr. Allen's critique may be clarified by three quotations:

1. Quoth about twenty distinguished geneticists attending the Eighth International Congress of Genetics: ". . . But it must also be understood that the effects of the bettered environment is not a direct one on the germ cells and that the Lamarckian doctrine is fallacious, according to which the children of parents who have had better opportunities for physical and mental development inherit these improvements, biologically, and according to which in consequence, the dominant classes and peoples would have become genetically superior to the underprivileged ones. The intrinsic (genetic) characteristics of any generation can be better than those of the preceding generation only as a result of some kind of *selection, i.e.,* by those persons of the preceding generation who had a better genetic equipment having produced more offspring, on the whole, than the rest, either through conscious choice, or as an automatic result of the way in which they lived. Under modern civilized conditions such selection is far less likely to be automatic than under primitive conditions, hence some kind of conscious guidance of selection is called for. To make this possible, however, the population must first appreciate the force of the above principles, and the social value which a wisely guided selection would have."

2. Quoth Sir Francis Galton: "Eugenics is the science which deals with all influences [under social control] that improve the inborn qualities of a race; also with those that develop them to the utmost advantage."

3. Quoth Alice as reported by Lewis Carroll: "But 'glory' doesn't mean 'a nice knockdown argument'," Alice objected. "When I use a word," Humpty-Dumpty said in a rather scornful tone, "It means just what I choose it to mean —neither more or less" . . . "The question is, which is to be master—that's all."

Dr. Allen is in error when he says that my perplexities regarding the Eugenic Hypothesis are "based on a misunderstanding." My problem is not *misunderstanding,* it is an *inability to comprehend*—very likely due to a genetic deficiency of some kind—the arguments outlined in his paragraphs 4, 5 and 6. "Deliberate environmental linkage" impresses me greatly—but *what does it mean?* How does it happen that in the *same environment* the "right people," genetically are linked to philoprogenitiveness by the environment, while the "un-right" people aren't? If the sheep and the goats are going to be picked out and processed in a different environment, then the proposal fails because deep down inside we know that such a manipulated society apes the fascists and the communists, and violates the concept of individual sanctity and freedom of choice.

Of course, birth selection and death selection aren't going to be the same thing. But whatever the process, selection has to be *effective.* It has to be effective in terms of over-all fitness as well as in terms of the characteristics to be modified. Either such

competent people as H. J. Muller and George Beadle are completely off the beam or elimination of selection for a character means the inevitable deterioration of the character. The rate is slow, no doubt, but it is not zero. This inevitable retrogression is a by-product of human action in changing the selection pattern. We created the problem. We can't make it go away by waving a verbal wand.

If we *could* figure out a way to put Galton's concept to work, effectively, the twenty-odd distinguished signers of the so-called "Geneticists Manifesto" opined that within not very many generations the dullest humans of that future time might rate as geniuses in our day. Maybe that would be a bad idea: just too much brilliance. Surely it would be a mixed blessing if all these geniuses had several hereditary diseases.

It is quite certain that an excess of geniuses—either whole or crippled—will never become a problem if we make eugenics a game of semantic musical chairs. Even Humpty-Dumpty can't pay his genes off when they work overtime.

The eugenic time-scale has to be cast in generations, but dare we be smug about the "retrogression in certain spheres"? It is a big undertaking, inventing something as effective as death selection, and less cruel and painful. I just wish I were *sure* how it is going to be done in an acceptable social context.

As the greying make-up man said to the editor who insisted that two more lines *must* be added to the page: "Lady, there's just no use arguing with a steel chase." I wish I were sure which of us is arguing with a steel chase. Maybe we both are.

ROBERT C. COOK
Director, Population Reference Bureau

Dr. Allen's Reply to Mr. Cook

I hope that Cook's good humor is an indication that further exchange of opinion will not impede constructive thought in any quarter.

I would myself endorse that part of the so-called "Geneticists' Manifesto" quoted by Cook provided that a carefully planned "Eugenic Environment" is included under the expression, "some kind of conscious guidance of selection." However, I believe that anybody who supposes that these twenty distinguished geneticists were in that statement denying anything that is asserted by the Eugenic Hypothesis either misunderstands or misinterprets the Eugenic Hypothesis. If Mr. Cook is unable to comprehend how *in the same environment* the "right people" would have more desire for children than the "un-right people," I can only urge him to study again Osborn's original discussion of the Eugenic Environment, because if it could be perfected that is exactly what it would accomplish.

Cook's "inability to comprehend" takes a rather damaging character with his suggestion that Osborn's proposed environment might resemble a fascist or communist system. The Eugenic Environment is to my knowledge the only serious and detailed proposal of positive eugenics that would be proof against discriminatory practices in the hands of an unprincipled government. For it depends on the availability *to everyone* of a wide variety of good environmental influences. These, however, would be selected by public assent so that those influences favoring fertility would be understood and appreciated mainly by the more intelligent parents. Thus the sheep *would separate themselves* from the goats by their own cultural responses. While Osborn has also urged a higher development of eugenic counseling, which tends to be authoritarian, this is only as an ancillary measure, and is not relevant to the Eugenic Hypothesis.

The quotation from Galton does surprise me, because in its second part it clearly leaves room within the meaning of the word, eugenics, for a program of birth selection aimed at improving the cultural heritage. Certainly the present inborn

qualities of a race cannot be developed to the utmost advantage until all children are raised in optimal home environments, and it therefore is within the scope of eugenics to learn what home environments may be optimal.

As for the third quotation, there is an obvious difference between changing the meaning of a word at will and reasserting the one and only meaning of a technical term such as Osborn's "Eugenic Environment."

It would be unfortunate if eugenists allowed this sort of debate to divide them. The Eugenic Hypothesis is advanced not as a basis for immediate social action, but as a basis for research. Much relevant research is already in progress, and these issues will eventually be settled by facts.

FERTILITY CONTROL IN THE LIGHT OF SOME RECENT CATHOLIC STATEMENTS

WILLIAM J. GIBBONS, S.J.
Loyola College, Baltimore

This is Part One of a two part article

THE PAST FEW YEARS have witnessed a definite sharpening of Catholic thought on the ends of marriage, and hence indirectly on the subject of fertility regulation. This has not been accomplished without considerable discussion within the Church, as well as sharp differences between Catholic writers and those advocating contraception as an appropriate means of alleviating economic and other pressures sometimes associated with high fertility. These discussions have for the most part been fruitful, especially in bringing to the fore grave moral and social issues involved and in drawing to the attention of many, certain emergent problems whose importance might not otherwise have been so quickly perceived.

This present paper makes no pretense of reviewing all contemporary Catholic literature on sexuality and the ends of marriage, nor does it aim at placing this literature in full historical perspective. What it does is focus attention on some authoritative statements of recent years, with a view to clarifying the basic Catholic position on fertility regulation. Admittedly, not all outside the Church share in detail the convictions of the Catholic authorities and theologians on this subject. For that matter, not every Catholic appreciates fully as yet the doctrinal implications and the supporting arguments of the latest decisions and judgments.[1] Nevertheless it is important that both the substantive content and the practical consequence of the teaching be properly understood. Hence a reporting paper like this one at the present time.

In the period following World War II the long-term and global consequences of accelerated rates of natural increase have been widely discussed. So too the more immediate problems arising from local and regional imbalances between population and resources, or between labor force and job opportunities. Along with increased economic activity in underdeveloped areas of growing population, on more than one occasion fertility regulation in various forms has been advocated and promoted. Now, the moral implications of such suggestions could not long escape the notice of Church authorities and theologians concerned with problems of human sex behavior. Quite apart from the merits of arguments put forth by those interpreting factual data in varying ways, it was clear a new and significant trend of thinking on human reproduction must hereafter be taken into account.

But this growing awareness of the population-resource relationship, and the problems presumably associated with it, was not the sole or even primary reason for certain of the Catholic moral judgments forthcoming on sexuality and marriage. Simultaneously with the mounting discussion of rapid population growth, other ideas, not necessarily connected with the social aspects of fertility, were being spread abroad by American and European authors. These had to do with the fundamental nature and purposes of sex and wedlock, and hence

[1] At times there seems to be question whether American writers on the subject of sexuality and marriage have had sufficient access to pertinent Roman documents and statements, if not in the original Latin or Italian at least in reliable translations. Gradually this problem is being resolved.

over time would have profound moral repercussions in personal and family living. This developing line of thought threatened, it would appear, to subvert traditional Christian teaching on sex and concupiscence and hence was not unrelated to dogmatic beliefs on the nature of man, original sin, and the effects of redemption. The growing literature on sexuality placed such emphasis on the ends of marriage other than reproduction, that the less educated especially might lose sight of nature's purpose in giving man sex, and the objective norms which should guide him in its use.

It is perhaps worth noting that the stress in recent papal teaching has been placed on the finality of sex and marriage as such,[2] on the proper ordering of acts which relate to procreation, on respect for the human life which comes into being with conception. The Church, moreover, in its official pronouncements has not endeavored to resolve economic and social problems in their technical details.[3] Nor has the Pope passed judgment on various demographic hypotheses and conclusions concerning the population-resource relationship in its strictly scientific aspects.[4] As a religious and moral teacher, he has, however, pointed out the moral limits within which practical action must be confined. At the same time he has urged that all reasonable efforts be made to improve the temporal lot of those in need, through increased productivity and better distribution, and that unnecessary restrictions be not placed between families and the means of sustenance.[5]

Within the past five years there have been a number of significant statements by the Pope on the subject of marriage and

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[2]By the finality of sex and marriage is understood the purpose intended by nature, and hence the Author of nature, in endowing man with reproductive capacity. The Oxford English Dictionary defines *finality* in its philosophical meaning, as the purpose or end of the thing caused. It relates finality to *final cause:* the end or purpose for which a thing is done, viewed as the cause of the act. Catholic teaching, adhering in this matter to the *philosophia perennis* (perennial philosophy), of which Aristotle and St. Thomas are leading proponents, insists that in evaluating the ultimate morality of a human action not only must the subjective attitude (including intent) of the actor be taken into account, but also the objective finality or purpose as intended by nature for the particular faculty or capacity being used. Thus to eat or drink for sociability or pleasure is not wrong in itself, but can become so if these ends are pursued entirely apart from, and perhaps to the detriment of health and physical well-being, the preservation of which is nature's purpose in giving man an appetite for food and drink.

[3]In *Quadragesimo Anno* (1931) Pope Pius XI thus clarified the relationship between strictly scientific and technical knowledge in the social field, and moral teaching and practice: "It is not of course the office of the Church to lead men to transient and perishable happiness only, but to that which is eternal. Indeed 'the Church believes that it would be wrong for her to interfere without just cause in such earthly concerns' [cf. Leo XIII, *Rerum Novarum,* para. 13]; but she never can relinquish her God-given task of interposing her authority not indeed in technical matters for which she has neither the equipment nor the mission, but in all those that have a bearing on moral conduct. For the deposit of faith entrusted to Us by God, and Our weighty office of propagating, interpreting and urging in season and out of season the entire moral law, demand that both social and economic questions be brought within Our supreme jurisdiction, in so far as they refer to moral issues." *Quadragesimo Anno: Encyclical Letter of Pius XI.* Washington, National Catholic Welfare Conference, 1940. p. 14. Original Latin in *Acta Apostolicae Sedis,* 23 (1931) 190. (The *Acta Apostolicae Sedis,* often abbreviated AAS, is the official commentary of the Holy See and is published periodically by the Vatican Polyglot Press. In content and form it is analogous to the *Federal Register* of the U. S. government.)

[4]Cf. Pius XII, Address to delegates to World Population Conference, September 8, 1954. English text in *Catholic Mind,* 53:256, April, 1955.

[5]Cf. Pius XII, Radio message of Pentecost, June 1, 1941. *Acta Apostolicae Sedis* 33 (1941) 216-227 (English version); *American Ecclesiastical Review,* 105:276-88, October, 1941.

[6]The Sacred Congregation of the Holy Office was founded by Paul III in 1542 to combat errors of doctrine. Its principal function as presently constituted is to safeguard the integrity of faith and morals by examining and evaluating ideas and statements of a religious and moral nature which are both significant and capable of propagating or leading to doctrinal error. Its Prefect or head, unlike some other Congregations, is not a Cardinal but the Pope himself. The Holy Office issues its decrees and warnings (*monita*)—which are far fewer than some believe—after careful study by a commission of Cardinals. It carries on its day to day operations under the direction of an Assessor, assisted by a sizable panel of consultors which includes certain Vatican officials and a number of professional theologians with varying backgrounds and specialties. In modern times especially, the Holy Office has shown great care in not going beyond the precise doctrinal point at issue, thus allowing the maximum of legitimate discussion and difference of opinion among qualified writers and theologians. When it does speak, it is with the full authority of the Holy See itself.

sexuality, as well as several pertinent decrees of the Holy Office.[6] Prominent among the papal statements were the encyclical letter on virginity,[7] the address to the Italian midwives,[8] and the address to the Family Front.[9] The Holy Office found it necessary to issue a warning (monitum) concerning certain types of literature on the sexual aspects of marriage, and especially those advocating *amplexus reservatus*.[10] It also condemned and placed on the Index the work of a noted French Catholic writer[11] on problems of sex, presumably because its treatment of sex morality seemed to admit excessively subjective norms and hence might be interpreted as tolerant of morally dangerous practices.[12] Through such statements as those indicated, the Holy See endeavored to correct tendentious and erroneous views on sex which have become common in the post-war years. The doctrinal points made in these statements will be touched upon in the course of the following discussion.

I. *Empirical Science and Ethical Values*

Basically there should be no serious disagreement between the Church and modern science, since all truth, natural and revealed, has the same God of Truth for its author. In principle, any misunderstandings which would arise, because for example a hypothesis is asserted as a certain conclusion, can be resolved as all concerned come to realize the facts, and as theory is brought more in line with reality objectively discernible. In practice, however, the resolution of disagreements does not always proceed smoothly. It may at times be unduly retarded because some spokesman for religion fails to distinguish sharply enough between clearcut doctrinal and moral teaching, and his own personal judgments. Or it may be, and not infrequently is, because some scientist transgresses the bounds of his specialty and attempts the role of moral teacher for which he may not be particularly qualified.[13]

The rights and needs of scientific enquiry carried on in a manner consonant with morality, have long been recognized by the Church. In centuries past, popes and many prelates showed special interest by acting as patrons of the arts and sciences. In more recent years, outstanding scientific merit has received due recognition through the Pontifical Academy of Sciences.[14] Moreover, practically every year since World War II, Pope Pius XII received one or more scientific congresses or their delegates in papal audience, usually taking the occasion to pay understanding tribute to their respective disciplines. That this solicitude of the Holy See is not confined to the

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[7]Pius XII, *Encyclical "Sacra Virginitas" on Holy Virginity* (unofficial translation). Rome, Vatican Polyglot Press, 1954; also, *Catholic Mind*, 52:491-512. Latin text in *Acta Apostolicae Sedis*, 46 (1954) 161-91.

[8]Allocation of Pope Pius XII on October 29, 1951 to convention delegates of Italian Union of Midwives. Italian text: *Acta Apostolicae Sedis*, 43 (1951) 835-54. A Latin translation, with informative annotations and documentary citations, of both this and the subsequent address to the Family Front, will be found in *De Re Matrimoniali*, by Francis Huerth, S.J. Rome, Gregorian University Press, 1955. Pertinent excerpts in English, with annotations, are in T. L. Bouscaren, *Canon Law Digest*. Milwaukee, Bruce. 1954. Vol. III, *passim*.

[9]Allocution of Pope Pius XII on November 26, 1951 to members of the Family Front. Italian text: *Acta Apostolicae Sedis*, 43 (1951) 855-60.

[10]Monitum of the Holy Office, June 30, 1952; *Acta Apostolicae Sedis*, 44 (1952) 546. English translation: Bouscaren, *op. cit.*, III, 435.

[11]Marc Oraison, S.T.D., M.D., *Vie chrétienne et problèmes de la sexualité*. Paris, J. Lithielleux, 1952.

[12]Decree of the Holy Office, March 19, 1953. *Acta Apostolicae Sedis*, 47 (1955) 46. Publication of the decree was delayed until January 3, 1955, by which time it was evident ideas the book contained were spreading and being quoted in other French works on the subject. The author, a competent writer known for concern about doctrinal integrity, immediately indicated his submission to the decree of the Holy Office. Notification of this fact was made in *Acta Apostolicae Sedis*, 47:89, February 25, 1955.

[13]Cf. Gibbons, W. J., "Catholic Value System in Relation to Human Fertility," in *Studies in Population* (ed. by George Mair). Princeton Univ. Press, 1949.

[14]Founded in Rome in 1603, and subsequently reorganized by Pius IX and Leo XIII, it was reconstituted in 1936 by Pius XI. A list of its officers and members appears in the *Annuario Pontificio* (Rome: Vatican Polyglot Press).

natural sciences, but extends as well to the medical, psychological, and social sciences, may be inferred readily from the list of groups received and messages or addresses delivered by the Pope.[15]

The Pope, needless to say, cannot guarantee that the same sympathetic understanding of science will be manifest everywhere and always among Catholics. There are admittedly a number of, perhaps too many, Catholics who adopt an anti-scientific attitude and instinctively suspect the findings of empirical science. This is regrettable, and clearly out of accord with the love of truth, whatever its source, which should characterize the dedicated Christian. But it must be recognized that a human variable, not necessarily connected with religion as such, is here involved and that cultural and socio-economic background of particular Catholics may be the underlying reasons for certain of their reactions in the face of scientific fact and theory. Looking at the matter in the long view of history, science by and large has fared better in Catholic surroundings than among some other groups.

There is of course another side of the picture. Whatever the shortcomings of individual Catholics, the Church as such has the mission of safeguarding moral truth from human distortion and in line with that duty has a unique teaching authority in these matters. In carrying out this mission, conflict does not ordinarily occur when it is simply a question of reconciling revealed truths and traditional ethical teaching with ascertained facts and soundly constructed scientific theory.[16] Misunderstandings between social scientists and social leaders on the one hand, and theologians and churchmen on the other, is much more apt to occur when action programs are involved. Then there is a likelihood the human behavior patterns fostered or proposed may impinge directly on the moral order of which the Church by definition is custodian. In attempting to resolve the conflicting views, the official Church tries to allow the maximum of leeway to conflicting views and to varying interpretations of empirical facts as these relate to human behavior. But there comes a point at which it has no choice but to insist on basic moral principles, emphasizing that however legitimate the objectives only morally acceptable means can be used in pursuit of them. The Church of course is under no illusion that men do or will adhere universally to the norms of conduct proposed—her whole mission in the present order presupposes the reality of sin—but she must do what is in her power to see that the norms themselves are not lost sight of through confused thinking or human weakness and self-deception.

II. *The Ends of Marriage and Fertility Regulation*

In historical perspective, it is not surprising that a developing body of thought emphasizing ends of marriage other than reproduction, should have such an impact on modern society. The industrial revolution, in the context of the larger scientific revolution of which it was a part, had paved the way for a new era as regards population growth and also popular attitudes toward family size. Especially with the advent of the nineteenth century, overall death rates declined rapidly, and with them rates of infant mortality. It was not long before spouses perceived rather generally the implications of this trend in relation to size of completed family and number of children ever born. They saw possible complications for themselves, even when not averting formally to broader social import. Other things being equal, surviving children hereafter were likely to be more numerous than in earlier times when high mortality more nearly balanced high

[15]Cf. *Catholic Periodical Index*, under heading "Pius XII" for list of addresses and groups to which given; also, location of original and English texts.

[16]Cf. Encyclical Letter *Humani Generis* (1950) on importance of effecting an integration of revealed truth and natural knowledge. *Acta Apostolicae Sedis* 42 (1950) 561-78; *Catholic Mind*, 48:688-700, Nov., 1950.

fertility. Finally, in the changing socio-economic structure of urbanized society, large family size was coming to be regarded by many as an obstacle to the improved levels of living made possible by technological advance.

In the concrete, then, there had emerged strong economic and psychological pressures on spouses to limit offspring. This intensified motivation for fertility regulation was accompanied by greatly increased accessibility to contraceptive information and materials. Promotional movements, demanding legal acceptance or at least toleration of the new techniques, came into being. Among both theoretical writers and social welfare leaders, there developed a tendency of speaking about marriage in such a way as to encourage effective separation of reproductive function and routine sexual performance. Modern civilization thus becomes characterized by a pro-contraceptive outlook. Nevertheless, rates of natural increase failed to decline as rapidly as anticipated by some, and in a number of areas actually accelerated, chiefly because death rates declined more rapidly than did fertility.

Now the desire and effort to dissociate sexual expression and childbearing, except as voluntarily determined, has existed for centuries, as such longstanding practices as withdrawal, abortion, and infanticide clearly indicate. That these practices were less common in Western civilization than in other times and places, may in large measure be attributed to the restraining influence of Christian ethical norms. To the extent such influence has been weakened by decline in religious belief and behavior, these practices have tended to spread. Yet Western man generally speaking, still regards infanticide and abortion as much more objectionable morally than other forms of fertility regulation. Consequently, there exists a psychological block against them. This fact, in combination with mounting pressures for family limitation, presumably accounts for the prevailing cultural preference for contraception referred to above.

Even those whose consciences would not permit reconciliation with certain of the popularized methods, became aware of possibilities for fertility regulation through systematic use of the female sterile period. For the Church and its leadership, this latter development posed additional problems of a moral and pastoral nature, over and above those initiated by widespread recourse to artificial contraception.[17] Although differing radically from non-natural methods of forestalling conception, use of the rhythm as it came to be known, was not unrelated to the general trend toward separation in thought and practice of sexual gratification from reproductive consequences. Evidently, then, a need had arisen for continuing clarification of Christian thought on the whole subject of sexuality and the ends of marriage. Church authorities, it may be noted, were quick to perceive this need, as may be gathered from the timely publication of the encyclical *Casti Connubii* by Pius XI in 1930.[18] They also realized the need would not be met by ignoring family and demographic problems, nor by giving in to the demands of the "new morality" as it was called by Pius XII in an address to the convention delegates of the World Federation of Catholic

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[17]The Sacred Penitentiary (in accord with its function of handling cases of conscience) in a reply of June 16, 1880 to the question: Is it licit to use marriage only on those days when conception is less likely, indicated that spouses who acted thus were not to be disturbed in conscience, especially since this practice was morally preferable to onanistic contraception. On July 20, 1932, to a new question: "Is that practice of spouses licit, whereby for good and serious reasons wishing to avoid offspring legitimately, by mutual consent and with proper intent they abstain from the use of marriage except on those days when according to the latest theories, conception is unlikely for natural reasons," the Sacred Penitentiary replied: The question has already been handled in the reply of June 16, 1880. For Latin text, cf. Huerth, *De Re Matrimoniali*, p. 111.

[18]*Acta Apostolicae Sedis*, 22 (1930) 539-91. English text in *Social Wellsprings*, ed. by Joseph Husslein. Milwaukee, Bruce, 1942. Vol. II; *Catholic Mind*, 29:21-64, January 22, 1931.

Young Women.[19] In fact, they could not do the latter in view of their obligation to interpret the principles of objective natural law morality, as controlling and being superior to problems of personal values and the ethics of situation.[20]

Traditionally, the ends of marriage have been formulated as to include the procreation and education of children, and the advantages accruing to the spouses from communal bed and life. These latter are understood to embrace the alleviating (remedying) of concupiscence (sex urge), as well as what other benefits may come from dwelling under the same roof and sharing each other's burdens and difficulties. In recent years, increasing stress has been laid by some authors on the so-called psychological complementarity of the sexes, and on the personality development said to occur in conjugal living with regularized sex expression. Provided such "complementarity" is understood properly, and not as justifying a reversal of the ends of marriage as given by nature, it surely merits consideration and further study. But if it means that only thus can human personality develop satisfactorily, or that reproduction may be regarded as a secondary and incidental value in marriage, then it implies a definite departure from tradition and natural law concepts.

Catholic teaching, as expressed in the Code of Canon Law, has formulated the concept thus: "The primary end of marriage is the procreation and education of offspring; the secondary end, mutual aid and the remedying of concupiscence."[21] The marriage contract, moreover, is understood to be essentially monogamous (between one man and one woman) and indissoluble. This latter precludes successive marriages so long as a previous spouse remains alive. Finally, Christian marriage is necessarily a sacrament, and thus attains a special firmness or solidity.[22]

Throughout history the Church has found it necessary to enunciate repeatedly these ends of marriage, defending the institution against those who would disparage it or else relax its God-given norms.[23] Some there have been who would restrict its use solely to the procreation of children. Others have emphasized to the point of exaggeration the secondary and subordinate ends. Both these tendencies the Church resisted, and today, while making due allowance for new insights reconcilable with the traditional moral framework, will continue to resist.

Marriage as such was disparaged by certain heretics of the early Christian era. These on occasion proposed very rigid norms, forbidding second marriage entirely, calling in question the goodness of even first marriage, or perhaps characterizing the institution as a lesser evil permitted by God but not one to be praised. Thus, varying aberrations from tradition are found among the Manicheans, the Montanists, and in the later Tertullian.[24] Even certain of the Church Fathers, writing against those who equated marriage and virginity or who leaned in the direction of corrupt pagan attitudes, at times gave the impression of minimizing the basic goodness of chaste wedlock. Thus, perhaps St. Jerome in his *Adversus Jovinianum*. And some have interpreted Augustine as not sufficient-

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[19] *Acta Apostolicae Sedis*, 44 (1952) 413-9. English text: *Irish Ecclesiastical Record*, 78:137-42, August, 1952. For Italian and Latin texts, and also annotations, cf. *De Conscientia Christiana atque De "Morali Nova"* (Reprint from *Periodica*, 41 (1952) 183-249). Rome, Gregorian University Press, 1952.

[20] In the address referred to above, the Pope points out the problems created by, and the limitations of existentialist morality and of the "ethics of situation" when these come in conflict with the more objective norms of natural law morality.

[21] *Codex Iuris Canonici*, c. 1013, §1. Cf. *Code of Canon Law; Text and Commentary*, by T. L. Bouscaren and A. C. Ellis. Milwaukee, Bruce, 1946.

[22] *Ibid.*, c.1013, §2.

[23] On the opinions of the Fathers on marriage, as well as of early Christian writers generally, cf. F. Cayré, *Manual of Patrology and History of Theology*, trans. by H. Howitt. Tournai, Desclée, 1936-40. 2 vols. See also: G. H. Joyce, *Christian Marriage: An Historical and Doctrinal Study*, New York, Sheed & Ward, 1933.

[24] Cf. Tertullian, *Treatises on Marriage and Remarriage*, trans. by W. P. LeSaint, S.J. (Ancient Christian Writers, No. 13). Westminster, Md., The Newman Press, 1951.

ly appreciative of marriage, since he too found it necessary to oppose the disparagers of virginity and celibacy.[25]

But a careful reading of Augustine will dispel this impression and lead to the conclusion he understood in an acceptable manner the ends of marriage and their ordering among themselves.[26] Even if at times Augustine's precision of expression leaves something to be desired, it is evident he recognizes the use of marriage for secondary ends as legitimate, and under proper circumstances praiseworthy. What he insists upon is that nature gave man sex because the race needed propagating, and that even spouses may not act as if procreation were not the first purpose of the institution, the one giving it justification and finality.

In our day the pendulum of speculative thought has swung in a direction contrary to that of Augustine, at least among not a few non-Catholic writers, and also among some who profess Catholicism.[27] Not only are the psychological values and the personality-development aspects emphasized, but some seem to imply that in the psychological and physical complementarity of the spouses the meaning and end of marriage is to be found. Thus offspring may be regarded as secondary or perhaps incidental consequences of the union of souls and bodies in the conjugal state. Manifestly much is yet to be said in sociological and psychological literature about these psycho-social aspects of marriage, and research could be done with profit. But there was, and apparently still is, a danger that not only will terminology be corrupted, but thinking will become confused. Accordingly, in 1944 the Holy Office took special cognizance of the trend and issued a decree on the ends of marriage and the interrelations between these ends.[28] It stated in substance that the traditional terminology is to be adhered to, namely that procreation and education of children is the primary or basic end, and that all other ends are subordinate to this. It is therefore incorrect to assert that all ends are essentially of equal importance, or that the subordinate ends, in the order of nature, exist independently of the primary end. Returning to the same theme in his address to the midwives (1951) Pius XII warned against the exaggeration of "personal values" to the extent that the principal purpose of marriage is lost sight of.[29] He also took the occasion to condemn what he called "sexual hedonism," namely the inordinate pursuit of sexual pleasure and its exaltation to an autonomous status, often quite without regard for the reproductive aspects of married life.[30]

[25]Before he wrote on *Holy Virginity*, Augustine made clear his esteem for the married state in *De Bono Conjugali* (The Good of Marriage). For English texts of both these writings, see *Saint Augustine: Treatises on Marriage and Other Subjects*, trans. by C. T. Wilcox, *et al.* New York, Fathers of the Church, 1955.

[26]Cf. *De Bono Conjugali*, especially ch. 17 ff. In *De Sancta Virginitate* he warns those consecrated to a life of virtuous continence that because their state is in itself preferable, marriage is not therefore to be considered evil (ch. 18). One of the most striking passages of Augustine on marriage is to be found in Book XIV of *De Civitate Dei*. Here he discusses the problem posed by passion and concupiscence, indicating that marriage existed even before the fall, for the purpose of propagating the race, and that had not man abandoned the state of original justice, he would have retained control over his reproductive organs, using them for procreation as occasion demanded, but not being motivated by the powerful force of lust (cf. ch. 22-24). Saint Augustine, *The City of God*; Books VIII-XVI. New York, Fathers of the Church, 1952.

[27]In this connection, reference is usually made to several Catholic works of the pre-war and early war period, namely *Sinn und Zweck der Ehe*, by Prof. Herbert Doms (Breslau, 1935), translated into French as *Du sens et de la fin du mariage* (Paris, 1937), and *Die Zweckfrage der Ehe in neuer Beleuchtung*, by Dr. Bernardine Krempel (Zurich and Cologne, 1941). After careful examination, both these books were ordered withdrawn from circulation. There is a brief but incisive discussion of this question in Huerth, *De Re Matrimoniali*, p. 117-23, where he considers the passages of Pius XII's address to the midwives which have to do with "personal values" in relation to the primary end of marriage.

[28]Decree of the Holy Office, April 1, 1944. *Acta Apostolicae Sedis*, 36 (1944) 103. English translation in Bouscaren, *Canon Law Digest*, III, p. 401-2.

[29]Pius XII, Address to the Midwives, nos. 42-47.

[30]*Ibid.*, nos. 61-67. Huerth (*De Re Matrimoniali*, p. 127) points out that there is a relationship between "sexual hedonism" and the advocacy of *amplexus reservatus*, concerning which there was issued the warning of the Holy Office on June 30, 1952. Cf. Bouscaren, *op. cit.*, III, p. 435.

(This article will be concluded in the June issue.)

TREATMENT IN HEREDITARY DISEASES

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SUCCESSFUL THERAPY AND PREVENTION of diseases are the final and highest goals of all medical science. Eugenic considerations aim mostly at prevention, partly for larger groups as in the problem of sterilization in certain mental diseases, and partly for individuals as in eugenic marriage counseling. This stress on prevention in spite of all its difficulties is due to the unfortunate fact that many hereditary anomalies do not yield to any of our present therapeutic measures. We have no means to improve most genetic forms of mental deficiency, certain kinds of congenital blindness, dwarfism and grave skeletal anomalies, malformations of the kidneys, inborn errors of metabolism which may cause very serious diseases, and many other genetic disorders.

Not infrequently we meet with the opinion that hereditary defects by their very nature are incurable. It must be admitted that the fertilized ovum already contains the pathologic genes which find expression later—during the fetal period, at birth, or during postnatal life—and that it is not possible to change the structure of the chromosomes or to transform pathologic factors into normal ones. In this sense a strictly causal therapy of genetically determined anomalies or diseases is not feasible. Causal therapy cures a disease by abolishing the primary cause and is illustrated by the use of antibiotics to kill invading bacteria and thereby to relieve symptoms of infection.

Because such an ideal form of treatment is not possible for hereditary diseases, two equally erroneous though very divergent attitudes may arise. One is therapeutic nihilism which gives up any attempt at correction where the damage is caused by genetic factors. On the other hand, therapeutic enthusiasts, *e.g.* in psychiatry, are inclined to overlook what in certain diseases are established facts of inheritance and even to deny altogether that genetic factors play a role, reasoning that a disorder which responds at all to therapeutic measures cannot be caused by genetic factors.

Actually, the error of both these extreme points of view can be illustrated for any field of medicine. In the following paragraphs only such pathologic conditions will be discussed in whose etiology, according to our present-day knowledge, genetic factors play a role. To establish the presence of such factors, the human geneticist who naturally cannot make use of experiments for his studies has three main methods at his disposal: the study of pedigrees, the statistical evaluation of observations on familial occurrences of certain traits, and the twin method. The last method compares the frequency of concordance of the character in question in one-egg and two-egg twin pairs. Since the partners of one-egg pairs are genetically identical and those of two-egg pairs genetically different, a significantly higher concordance rate among one-egg twin partners speaks in favor of a genetic influence upon the trait under study. In certain conditions the etiologic role of genetic factors is of major importance inasmuch as almost all cases are genetically determined and environmental factors play only a very minor role, if any. In particular, in many rare anomalies which typically run in families, pedigree studies are sufficient to delineate the underlying genetic factors. In other conditions hereditary factors may be operative only in a part of the cases or a hereditary disposition may be only one of several factors

whose cooperation leads to the actual manifestation of the anomaly or disease. The genetic study of such conditions, in particular if they are not infrequent in the general population, requires systematic family and/or twin studies with statistical evaluation of the results. Here we cannot go into details concerning the pertinent observations for each example discussed. As stated above, our discussion will be confined to conditions for which sufficient material has been collected to reveal the causal role of genetic factors.*

Forms of Therapy

1. Anatomical Correction

We have already stated that a strictly causal therapy for genetic disorders is not possible. However, effective anatomical correction by surgery is almost as good as causal therapy for the affected individual. Many congenital malformations belong to this category, such as clubfoot, congenital dislocation of joints (chiefly of the hip), harelip and cleft palate, polydactyly (supernumerary fingers and/or toes), syndactylism (fusion of fingers). All these congenital deformities can be corrected almost completely by modern plastic and orthopedic surgery.

More serious malformations, frequently fatal in early life, can be corrected only by major operations. In congenital pyloric stenosis (narrowing of the lower end of the stomach) the ring-like muscle which controls the passage of food from the stomach to the intestines is overdeveloped. Instead of controlling, it obstructs the food passage. In severe cases such children would be doomed to early death without treatment. A comparatively simple operative method, however, provides a complete and permanent cure. In another congenital malformation of the digestive tract, the so-called megacolon or Hirschsprung's disease, the terminal large bowel is dilated, sometimes to an enormous degree. This anomaly is not fatal, but it causes an excessive constipation which does not respond to the usual therapeutic measures. Two surgeons (Swenson and Bill) perfected a technique for removal of the dilated part of the bowel which promptly relieves the constipation and all its consequences.

Still graver are the congenital malformations of the heart and great vessels. (1) One example is persistence of the arterial duct which during pre-natal life bypasses the non-functioning lungs, and which normally closes after birth. Its persistence during postnatal life causes an insufficient arterial blood supply to the cells of the body and an additional strain on the heart which may lead to heart failure. Surgical ligation and division of the duct relieves this strain and provides sufficient oxygen supply to the tissues of the whole body. In another complicated malformation of the heart, the so-called tetralogy of Fallot, the anomaly consists chiefly in a displacement of the origin of the aorta. This vessel which normally originates from the left heart chamber alone is displaced in such a manner that some blood from the right heart chamber also enters it. The right heart chamber, however, contains unoxygenated blood; when such blood reaches the aorta and the general circulation, this is readily recognized by the intense blueish discoloration of the cheeks, lips, ears, fingers, and toes, and justifies for these patients the term "blue babies." These children suffer from severe shortness of breath and cannot develop in a normal way. By the ingenious operative method of Blalock and Taussig, however, surgeons are able to correct to a large extent the abnormal circulatory conditions and to relieve the strain on the heart.

In discussing the possibilities of anatomical correction, we must not forget the good results of orthodontic measures. Abnormal position of teeth and malocclusion can be corrected. Dental caries and decay, even when on a hereditary basis, (2) can be suc-

*The literature numbers given in the text refer only to a few of the more important genetic studies on the pathologic conditions discussed.

cessfully treated by conservative methods.

2. **Medical Replacement Therapy.** Another form of treatment which in its favorable effects almost equals causal therapy is the medical replacement of physiologic agents, especially hormones where these are deficient. The outstanding example in this connection is the treatment of diabetes mellitus. (3) In this metabolic disorder the utilization and oxygenation of sugar by the tissues and sugar storage in the liver are disturbed. Consequently the sugar level in the blood rises; sugar passes through the filter of the kidney and is eliminated in the urine. Aside from the infrequent cases of "symptomatic diabetes" appearing in certain disorders of various endocrine glands, the great majority of diabetics belongs to the so-called pancreatic diabetes. The normal pancreas produces the hormone, insulin, which has a profound influence on sugar metabolism, promoting the utilization of sugar by the tissues, increasing its storage in the liver, and lowering the blood sugar. Removal of the pancreas in the animal or deficient output of insulin in man causes diabetes. Although according to modern concepts a more complex disorder of sugar metabolism is present in such patients, it is enough to say that they are suffering from an insulin insufficiency. This lack of insulin can be controlled very successfully in the vast majority of cases by insulin treatment. In the pre-insulin era the most dreaded complication was the so-called diabetic coma. This is a state of unconsciousness with forced breathing, grave disorder of the electrolyte (acid-base) balance, and high blood sugar which usually ended fatally before insulin was available. This and other complications can be avoided or brought under control by modern treatment, and even in severe cases patients can be kept active and in a satisfactory state of health.

Historically the first efficient hormonal replacement therapy was the use of thyroid extract in diminished activity or complete lack of the gland. The thyroid hormone has a profound influence on almost every cell of the body, increasing oxygen consumption, stimulating metabolic processes and promoting growth. It also stimulates the autonomic (involuntary) nervous system, the heartbeat, intestinal activity, brain waves as seen in the electro-encephalogram, and many other functions of the organism. It is to be expected that a diminished supply of such a hormone should cause striking somatic and mental changes. Mentally these patients become slow and dull: they lack interests and lose their memory. The bodily functions are slowed down, also. The face presents a very characteristic, puffy, featureless aspect; the hands and feet are also puffy. If the lack of hormone begins during childhood, considerable retardation of growth and mental deficiency are the consequences. If started in time and properly handled, the administration of thyroid extract gives excellent results both in adult and in infantile cases. Of course, the lack of thyroid hormone during the fetal period cannot be completely compensated later, and though individuals with congenital complete lack of the gland often remain retarded in their skeletal growth, even such patients may develop into normal adults with average intelligence if treatment is started early and is continued faithfully.

Similarly, so-called eunuchoidism in the male, a constitutional deficiency in the development and function of the testicles, is amenable to replacement therapy with testicular hormone. Sexual behavior and other symptoms are cured, although sterility persists.

Among other instances of hormonal deficiency brief mention should be made of two diseases. The non-tuberculous form of Addison's disease may be hereditary. It is a grave and formerly a fatal condition characterized by extreme weakness and lowering of blood pressure and blood sugar, brown discoloration of the skin, vomiting, diarrhea and other signs. This disease is due to an insufficient supply of hormone from the outer, cortical layer of the adrenal

glands located at the upper pole of each kidney. The second disease is "idiopathic tetany," characterized by painful spasms in hands and feet and by lowering of the concentration of calcium in the blood. This disease is due to insufficient secretion of the parathyroid hormone which is produced by four small glands located at the back of the thyroid gland. In both these diseases hormonal replacement therapy combined with other measures for the restitution of the disturbed chemical balance is highly effective.

Among the hereditary deficiencies which are amenable to replacement therapy is so-called pernicious anemia (Addison's anemia) (1) which is due to a lack of the "erythrocyte maturation principle," a factor which regulates the normal development of the red blood cells. The term "pernicious" is no longer justified in this form of anemia since we have learned that it may be kept well under control by treatment with liver or vitamin B_{12}.

If, on the other hand, a gland is hyperactive, producing too much of its specific secretion, two methods are available to reduce its function to normal. One is surgical reduction of its volume, the other is irradiation therapy which destroys some of the secreting cells. Both these methods in their effect come as near to causal therapy as the replacement treatment though a certain risk inherent to all surgery cannot be avoided. The most important example is thyroid overfunction in its two somewhat different forms, Graves' disease and toxic adenoma of the gland. While in the latter only the cells of a part of the gland, mostly of a pre-formed nodule, are overactive, in the former, more dangerous kind the entire gland is enlarged and in a state of overactivity. Large amounts of hormone are produced and poured into the bloodstream, to be distributed to the whole organism. Considerable loss of weight, acceleration of the heart rate, excessive nervous excitability, poor sleep, sweating, diarrhea, and in Graves' disease a protrusion of the eyeballs are the most important features of the clinical picture. In severe, untreated cases Graves' disease may be fatal or may do permanent damage to the heart. With the exception of a small minority of cases, the results of modern therapy are highly satisfactory.

3. **Surgery for Unfavorable Mechanical Conditions.** In another group of diseases not overfunction but unfavorable mechanical conditions can be corrected by surgical measures. Goiter without functional disorder of the gland which, however, compresses the trachea and hinders breathing, belongs in this category. Large fibromas of the uterus may cause difficulty of urination by pressure on the bladder or they may provoke pain by compression of nerves. The benefit of surgery in such conditions is evident.

4. **Other Therapy.** Unfortunately, in a great number of disorders complete cure is not possible by means of our present methods. However, we may be able to relieve symptoms, to avoid or delay complications, to stop at least temporarily the progress of the disease. Such therapeutic methods are admittedly far from causal therapy; however, they are decidedly beneficial and modern medicine could not do without them. Though we are unable to restore the eyesight of a person with cataract to normal, we still can prevent his becoming blind and enable him to see sufficiently well for the usual daily routine including reading and writing. There is a large number of hereditary diseases which belong to this group. In this connection we shall mention only a brief selection.

Among the diseases of the heart and vessels important in this connection is the most frequent form of permanently high blood pressure, so-called essential hypertension. Its incidence in the general population is considerable. Treatment is not very effective, but to a certain extent it relieves the patient's symptoms, delays disastrous complications, and slows down the progress of the disease.

In benign ulcer of the stomach (4) a defect in the inner lining of the organ penetrates to the muscular coat and causes pain and not infrequently bleeding. Very deep ulcers may cause perforation of the gastric wall and escape of the stomach contents into the abdominal cavity. Such a perforation is a very dangerous complication; it may be accompanied by shock and later followed by inflammation in the abdominal cavity. Constrictive scarring, furthermore, may lead to considerable narrowing of the farther end of the stomach with obstruction of the food passage. There is no doubt that medical and—where necessary—surgical treatment is very helpful in relieving the patient's distress, and in warding off complications such as those mentioned above.

Therapy is also fairly effective in some metabolic disorders like gout. In this disease the uric acid of the blood is increased. It is then deposited as a sodium salt in cartilage and in those parts of the bones which form joints. These deposits may cause either an acute attack of gout or chronic gouty arthritis.

Some blood diseases should be discussed in this connection. In the so-called congenital hemolytic jaundice the red blood cells have an increased fragility and therefore a shorter lifespan than normal ones. They are chiefly destroyed in the enlarged spleen. The patients have a more or less marked yellowish skin color through all their lives. In the course of the disease, periods of acute, outspoken anemia occur. Removal of the spleen has a favorable effect in this disease, in particular during the acute crises of aggravation and anemia. In "idiopathic thrombopenic purpura" a lack of blood platelets appearing without obvious cause is responsible for smaller or larger bleedings not only in the skin but also in inner organs. In more than half of these cases removal of the spleen is beneficial. True hemophilia was one of the first pathologic conditions where a hereditary pattern was studied in man. At present there seems to be well-founded hope for a successful therapy in the form of concentrated blood additives.

Among the various allergic disorders should be mentioned bronchial asthma, hayfever, vasomotor rhinitis (allergic oversecretion of nasal mucus), eczema and others. Although, of course, the allergic trait and tendency to recurrence remain, therapy is fairly successful in fighting the actual allergic ailment.

A few examples of diseases affecting the eyes and ears may be included here. In glaucoma the normal escape of intraocular fluid is blocked. Consequently the intraocular pressure rises, and this high tension leads to impairment of vision and finally to blindness. Senile or presenile cataract constitutes a progressive clouding of the lens of the eye with resultant impairment of vision. Treatment is fairly successful in averting blindness in both diseases. By means of a comparatively new surgical method, remarkable therapeutic results have been achieved in a number of cases of otosclerosis. In this disease bony changes within the inner ear produce progressive deafness.

Most complicated and difficult to judge are the effects of therapy in mental disorders. However, to a certain extent, modern therapy is successful in depressive states and in schizophrenia, where it often keeps patients functioning at a fairly satisfactory level for at least a limited, sometimes for an extended, period of time.

Last of all, cancer should be mentioned, which, of course, is amenable to treatment only in its early stages while complete removal of all cancerous tissue is feasible. Moreover, many authors doubt whether genetic factors play a part in its etiology at all. Other observers again are in favor of the assumption that genetic factors do play a role in cancer etiology, in particular that they are important in cancer of certain organs such as the female breast, the uterus, and the digestive tract.

We cannot conclude this list of treatable hereditary diseases without mentioning

that an ever-increasing number of studies suggest that some infectious diseases, notwithstanding their bacterial or viral etiology, are to a certain extent dependent on genetic factors, also. A higher susceptibility to the causal micro-organism may be an inherited trait. This has been demonstrated not only for chronic infectious diseases like tuberculosis but also for acute ones like poliomyelitis, tonsillitis, appendicitis and others. The recent great progress in the prophylaxis and treatment of these diseases is common knowledge.

The above-discussed examples are only a selection of treatable hereditary conditions and do not represent a complete survey. However, they do illustrate the fact that modern medical science has highly effective methods for dealing with many genetically determined disorders.

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FERTILITY IN MID-CENTURY AMERICA

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THE BIG NEWS IN HUMAN FERTILITY in mid-century America has been the 1940-1955 baby boom, which in dramatic significance has rivaled the hydrogen bomb explosions set off by the United States and Russia and the international atoms-for-peace movement that climaxed recently in a Geneva conference of the world's leading physicists. This increase in the U. S. birth rate upset not only predictions of demographers but also research interests and methods used for more than a generation. Before errors of prediction had become apparent, however, demographers had been aware of shortcomings and had been busy trying to elaborate theory and refine tools of measurement. Experience and insights gained from the Indianapolis Study, for example, contributed heavily toward better understanding of the factors underlying human fertility and toward new research orientations that would be more effective than old ones in defining and measuring these factors.

There is now keen interest among some students of human fertility in the ways in which current or proposed research has been shaped by the results of that study. We tend to expect of its successors a combination of the best theory and methods in projects of sufficient scope to shed further light on the basic factors that influence the having or not having of children. What actually is happening along this line can perhaps best be judged at this time by a critical review of the research proposal of the Milbank Memorial Fund for a new study of fertility and of the Puerto Rico research of Hill, Back, and Stycos. Our discussion first will summarize briefly what has been done on each of these two projects and then indicate some of the features of the work which deserve further consideration by the research teams.

The article by Mishler and Westoff on "A Proposal For Research on Social and Psychological Factors Affecting Fertility: Concepts and Hypotheses"[1] is a progress report by the technical staff of the Milbank Memorial Fund's Steering Committee on the development of plans for a new study of fertility. The objective of this study is basic research at the level of motivation in family planning and family size. Attention of the committee so far has been focused on problems of conceptualization and possible organizing principles. Research will be directed to knowledge of factors affecting successive orders of pregnancy or birth in the belief that such information is essential to an understanding of the size of completed families. According to present plans, the study will be limited to women who have had two children. Repeat interviews of these women 18 months after the first visit may be included in the research design in order to test the performance of couples over a specified period of time.

The conceptual framework for fertility research as outlined by the authors defines the pattern of contraceptive use as the major variable for study. Pregnancy desires and capacity to carry out these desires are more specific aspects of the pattern that are proposed for study. Factors related to

[1]Milbank Memorial Fund, *Current Research in Human Fertility*, "A Proposal For Research on Social Psychological Factors Affecting Fertility: Concepts and Hypotheses" by Elliot G. Mishler and Charles F. Westoff, pp. 121-150, New York, 1955. See also EUGENIC QUARTERLY, Dec. 1955, pp. 229-233.

contraceptive use, or the independent variables, are assumed to be the major elements of the concrete situation within which the fertility event takes place. These elements are considered on theoretically distinct levels of the person, the family group, and the social setting.

With the person as the unit of analysis, attention is directed to general personal attributes, such as excessive dependency needs, and to extra-familial orientations and roles that are incompatible with children. With the family group as the unit of analysis, styles of life and systems of organization are distinguished. Important styles of life are defined as familistic and work-mobility. Systems of organization are considered from the point of view of interaction patterns and psychological aspects. Power, communication, and division of labor are features of interaction patterns deemed useful for study while cohesiveness, marital adjustment, and norms constitute psychological aspects of importance. The social setting is viewed as characteristics of other social groups that are more or less compatible with various patterns of fertility performance. Continuity of membership and status in social groups and the benevolence of the psychological, social, and economic environment provided by these groups are selected as areas of the social setting that should be investigated.

The operation of this theoretical model is taken to be dependent upon the extent to which having a child or a certain number of children is compatible with other life values and interests. "Although at the level of a truism this serves to bring into prominence the notion that a fertility choice (whether implicit or explicit) involves a 'cost' for the individual and for the family, that is, certain desires and interests are either yielded or compromised in preference to others. . . . If it is given its widest and most general interpretation then all specific hypotheses and findings are simply manifestations or instances of this 'law' of compatibility. . . ."[2]

The authors have derived from this theoretical statement some 21 hypotheses that are grouped under the most suitable unit of analysis. Some of these hypotheses are broken down into sub-hypotheses with the fullest development made of those that pertain to socio-economic variables. The treatment is not complete, however, for the writers warn us that important issues and problems have been sidestepped or ignored and that hypotheses have been stated without necessary qualification and specification and have not been rated as to relative importance.

In a critical review of the work of the Committee, Hauser observes that we now have some theory and some specific hypotheses, but that there is yet to come a second stage of sampling, statistical and operational design and a third stage of design for testing the hypotheses.[3] His critique of the hypotheses is given under the headings of inclusiveness, operational form, and interaction of the variables. Under inclusiveness, he notes omission of "face-sheet" items, failure to take into account the timing aspect of the variables, lack of specific reference to the general social-economic climate, and shortcomings of fertility research that deals with less than the whole procreative period of the couples. Under operational form, he suggests the desirability of breaking down the dependent variable of psychological availability of contraception into its components, he points out that the "person as a unit" involves two spouses and that it is necessary to take into account their interaction, he raises the question of how many of the attributes or variables included in the hypotheses can be actually identified and measured effectively for purposes of testing the hypotheses, and he wonders whether the separation in time between acquisition of data and fertility performance is potentially great enough

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[2] *Ibid.*, p. 132.

[3] *Ibid.*, "Some Observations on **Method and Study Design**" by Philip M. Hauser, pp. **151-162.**

to make prediction useful. Under interaction of the variables, he points out that the proposal includes practically nothing on this subject but that the final study design must make some attempt to deal with the problem.

Our criticism of the research proposal must be conditioned not only by the tentative and skeletal character of the theoretical model in its present stage of development, but also by the fact that this area of research has not been sufficiently well staked off by prior studies to permit at this time a fully integrated and theoretically adequate investigation. Without pointing out obviously loose ends of the conceptual framework or marking the absence of qualification and specification noted by the technical staff itself, we believe that it might be helpful to indicate some of the danger areas that should be skirted as the research proposal is further refined and focused.

The proposal as now defined emphasizes the influence of a "work-mobility" ethic upon fertility and in so doing shows some cultural bias. The model, in fact, tends to be oriented principally toward middle class, urban, Protestant, and industrial society. This focus is very likely the result of leaning heavily upon the findings of the Indianapolis Study. There can be no quarrel with slanting a new study in the same direction. We advocate only explicit recognition of the kind of value net that is being spread to catch relevant motivational data.

A second possible bias is that of rational behavior in the "cost" theory of fertility decisions. Emphasis upon this theme is reminiscent of classical economics. This element, no doubt, is important for many couples in planning the size of their family and in carrying out their plans in the specific social and economic circumstances of early married life. There are many other couples, however, for whom this approach is not likely to yield much insight into the nature of fertility behavior. It is well known that planning is often only rationalization of deep-seated, unconscious social and psychological backgrounds. In families, furthermore, in which a strictly utilitarian approach is taken toward having children, we might well ask to what extent the positive and negative values of cost analysis are known. It is recognized in this connection that the couple's definition of the situation might be the important research goal.

With a cultural bias and a rational approach to fertility behavior, the authors have not neglected completely areas of interest that are pertinent to the field of psychoanalysis. The concepts of excessive dependency needs and of basic ego identification with parent of the same sex appear somewhat baldly placed in the total structure of hypotheses. The meaning of these variables is difficult to determine without ascertaining the matrix of other personal characteristics and the social setting that tends to direct, fulfill, or frustrate the need. Thus, the expression and meaning of specific personality traits depend upon the total operational constellation. How desirable is it, therefore, to open such areas of interest when their exploration in an effective way might consume the entire resources of the project? The staff has probably given consideration to this point and might have some good plan for its practical solution. Judging only from their statement, however, we see the possibility of superficial fishing in deep waters.

The whole field of motivation requires treatment that presents a dilemma to any research group with limited resources. The proposal of the technical staff already has exceeded the budget of the Committee and restrictions have been placed on the outline as presented in 1954 to the Milbank Conference. As conceptualization leads to further specification, additional tailoring may become necessary. In a relatively undeveloped field of research, the more adequate the theoretical formulation of a new study the greater the inability of the staff

to carry out the study of the problems that are deemed crucial in the whole structure of posited interrelationships. This is forever the dilemma of good research in the social sciences.

Another inheritor of the Indianapolis Study legacy is the Puerto Rico project, which has been underway since 1951 and from which results have begun to flow.[4] The primary purpose of this research was to discover why the actual family size as late as 1950 was about twice as large as the ideal size family of about three children. The authors began their study with the hypothesis that general motivations to take action with regard to family size, knowledge of means of control, and efficient family organization determine the relation between ideal and actual number of children. In the Milbank Conference paper, they described source, size, and composition of samples; outlined an analytical model of selected variables in fertility planning; stated hypotheses relating family action potentials to fertility control; and presented the results of an application of these hypotheses to a sample of 888 Island families. These couples met the criteria of husband and wife living together, married five to 20 years, of demonstrated fertility, and of less than sixth-grade education.

Hypotheses were classified under the following headings: (1) effectiveness in intra-family communication, (2) female modesty barriers to communication and action on fertility control, (3) empathy between spouses and action on fertility control, (4) marital satisfaction and action on fertility control, and (5) family organization. Under the last heading, three familistic types were distinguished on the basis of freedom of the wife to work gainfully, freedom from husband dominance, and freedom from prohibitions on her life space. Among the findings were: (1) degree of communication differentiates successfully among families on birth control use, but not on success with which used, (2) wives who correctly perceive husband's attitudes on having more children practice birth control more than those wives who less correctly understand their husband's attitudes, (3) marital happiness correlates well with general communication, but is indifferently related to fertility control, (4) marital agreement appears significantly related to success in fertility control, (5) wives with least satisfaction in the sex relationship are the most likely to have practiced birth control over a long period if they have practiced it at all, and (6) familism appears closely related to fertility, fertility control, and success in fertility control.

In this article, the authors unfortunately were unable to present much of the data they have obtained from their extensive research in Puerto Rico. Questions about representativeness of the sample, definition and measurement of variables, interviewing procedures, responses of persons interviewed, reliability of their answers, processing of schedules, and techniques of summarizing and relating the data cannot be answered from the limited information given in this paper. Although the brief description of what has been done indicates high-level research, it is not sufficient for a critical review of the work. Evaluation of the scientific character of the project, therefore, must await publication of their book in which they probably will give full details of methods used.

The data of the article, nevertheless, are adequate to raise some questions about the method used to test or prove the hypotheses relating efficiency of family organization to fertility. In Table 2, we are shown which of 36 zero-order correlations met the 5 per cent test of statistical significance. Twenty-one came up to this level and the hypotheses were accepted as supported. In the other 15 cases, the test was not met and the hypotheses were either rejected or reversed.

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4 *Ibid.*, "Family Action Potentials and Fertility Planning in Puerto Rico" by Reuben Hill, Kurt Back, and J. Mayone Stycos, pp. 42-62.

From such evidence, there is difficulty in knowing exactly what has been demonstrated. First of all, the approach is atomistic; whereas, the analytical model that is the source of the hypotheses is patterned. Unless all the items of the integrated theoretical structure are supported, we need to know whether the pattern as a whole falls, whether a major job of reweaving is required, or whether a patch-work of re-interpretation will mend sufficiently the structure for it to continue as an explanation of fertility. Secondly, tests of statistical significance tell us practically nothing per se about how much of the variation in fertility has been accounted for by the hypotheses. Thirdly, the general character of the independent variables, such as marital adjustment and familistic type of organization, might conceal other factors related to these variables that are responsible for their correlations with use of birth control and success in its use. Finally, even if correlations of high order were found and theoretical inconsistencies between supported and unsupported hypotheses were ironed out, the authors still would have the task of explaining the processes by which operation of the model yielded the correlations. Correlations need to be buttressed by qualitative materials and logical connections that indicate the processes through which the independent variables are associated with fertility.

Physical scientists are more accustomed than social scientists to so finishing the job of research and not letting it stop with the obtaining of statistical association between the variables. In the physical sciences, of course, possession of more closely-reasoned theoretical models that are supported by much past research of an additive character makes more possible the crucial experiment from which results can be taken directly and fed back into the analytical structure; in other words, the explanation of quantitative associations found in the test are at hand before the test is made. In the social sciences, on the other hand, it is necessary to backtrack from the correlations to other research and to the conceptual framework in order to juggle the pieces together in a new whole. Only as other parts of the model used in the Puerto Rican research are specified, hypotheses stated, and findings given will we know what amount of juggling would be necessary to fit these materials on efficiency of family organization into an adequate conceptualization of the whole process of fertility behavior. Only then will we know how much emphasis to place upon this factor of family organization as an explanation of the difference between ideal and actual family size.

So much for some of the newer research in social demography that has been rooted in soil prepared by the Indianapolis Study. Let us turn now to a consideration of current or proposed research that stems more directly from interests, traditions, and data of formal demography. We particularly have in mind studies making use of vital statistics, census reports, or related materials. The objectives of these studies are usually much more limited than those discussed above. Refined statistical analysis rather than elaborate conceptualization is the keynote or identifying characteristic of this second group of investigations. P. K. Whelpton's monograph on cohort fertility can be considered appropriately under this heading.[5]

The objective of his study may be considered as the accurate prediction of population trends, using past and present marriage and birth rates to estimate number of children to be born during the reproductive history of the female cohorts under 45 years of age included in a population at a particular time. One of the technical problems confronted by the analyst after breaking down the universe of women into subgroups homogeneous for age, parity, and marital status is that of removing cyclical influences from their past birth perform-

[5]P. K. Whelpton, *Cohort Fertility*, Princenton University Press, 1954.

ance. Surpluses and deficits of births resulting from a smoothing of the record are allocated to the proper time period in the reproductive history of each cohort. Thus, the timing of births with its postponements and accelerations under the influence of social and economic conditions is taken into account. Such refinement of the record of births, however, still leaves the problem of making projections. Accuracy of these projections depends upon correct estimates of the marriage rate, of the course of social and economic history as it might influence timing of births, of fashions in child bearing, and of secular trends in the cultural definition of the ideal-sized family. One way to improve these estimates might be to make them for cohort subgroups that are specific for residence, class, race, religion, and ethnic background as well as for age, parity, and marital status. How possible or practicable such a breakdown would be remains to be seen. A second method of acquiring information and insight needed for improvement of our ability to predict trends in birth rates is that of periodically surveying national samples of women for data on their past birth performance, plans for the future, and general attitudes toward child bearing and rearing.

Such a survey by Whelpton, Freedman, and others that is now in progress is a cooperative undertaking by the Survey Research Center of the University of Michigan and the Scripps Foundation of Miami University.[6] Their sample was to consist of about 2,700 married white women between the ages of 18 and 39 with husband present or in the armed forces and about 300 single white women between the ages of 18 and 24. These women were to be interviewed for approximately 60 minutes each by trained women interviewers on the staff of the Survey Research Center. Single women were to be asked about plans to marry, engagements, and ideal family size. Married women were to be asked about pregnancy histories, birth-control methods, and ideal family size. General background information was to be obtained from both groups. Results will be compared with actual births during the 1955-1959 period as registered with the National Office of Vital Statistics and as shown by the 1960 Census data on children ever born. It is hoped that similar surveys can be undertaken in 1960, 1965, and later years. Besides such comparisons of expected with actual births, valuable information will be obtained on sterility, family planning, birth-control methods, and differentials in fertility. The crucial problem faced by the study is how close a relationship there is between what people say they are going to do and what they actually do. No doubt, major discrepancies will be found. Success in using such material will likely occur as further surveys indicate circumstances that deflect or change opinions, desires, and plans of people with regard to having or not having children.

A third approach that might lead to improved predictions of cohort fertility is the more thorough analysis of the past response of marriage and birth rates to historical factors. N. B. Ryder's study of Swedish female birth cohorts from 1751 to 1901 is an example of what can be done along this line when excellent data are available.[7] The author states that his material may be summarized as (1) the challenge to a population posed by a rapid increase in survivorship; (2) the initial response to the problem by a stringent limitation of marriage; (3) the frustration of this particular means of family limitation; and (4) ways in which the above demographic developments were manifested in family life. According to Ryder, the most striking result of the investigation is that it suggests the probability that Swedish fertility began declining per-

[6]Milbank Memorial Fund, *Current Research in Human Fertility*, "A Study of the 'Expected' Completed Fertility of a National Sample of White Women," by P. K. Whelpton, pp. 106-112.

[7]*Ibid.*, "The Influence of Declining Mortality on Swedish Reproductivity" by N. B. Ryder, pp. 65-81.

ceptibly not in the latter part of the nineteenth century, but rather some 70 years earlier. He concludes: ". . . The following propositions may be asserted with a fair degree of confidence: (1) There was a marked decline in nuptiality of the Swedish female population, beginning early in the nineteenth century; (2) although this change induced some decline in fertility the effects were practically nullified by contemporaneous changes in mortality; (3) the major decline in fertility awaited the abandonment of what Malthus called 'moral restraint' in favor of the Neo-Malthusian remedy for excess fertility."[8]

These conclusions arise out of a preliminary report with 14 charts showing mainly changes from 1751 to 1901 in total person years, person-years married, nuptiality rates, mean marital duration, female live births, gross fertility rates, and marital fertility rates. On the whole, the data have been handled carefully in their graphic presentation and clearly indicate the course of several measures of fertility over 150 years as mortality declined. The author shows awareness of technical problems that confront the student in measuring fertility changes. His interpretation of the data tends to be forceful and dramatic.

His main weakness as revealed by this preliminary report appears to be over emphasis on the teleological adjustment of Swedish cohorts to declining mortality. In order to bring out this character of his interpretation, we present below some of the facts that, for the most part, are only implicit in the data of his graphs.[9] From 1751 to 1851, the number married in the cohorts of 100,000 increased from about 47,400 to 55,000. The number of children per 1,000 married person-years remained fairly constant at around 230 for the hundred-year period. The average age of marriage also was stable, being approximately 27 years. All of these facts suggest no adjustment to the 30 per cent increase during the same period in total person-years, ages 15-50, for the female cohorts of 100,000. What did change was the proportion of females who married. In the cohort of 1851, for example, there were approximately 14,000 more females who arrived at a marriageable age due to improvement in mortality during the preceding 100 years. If the nuptiality rate had remained the same as for the 1751 cohort, about 11,250 instead of 7,600 of these 14,000 would have married. Thus, for every cohort of 100,000 near the middle of the nineteenth century there were about 3,500 females who remained single who would have married if the nuptiality rate of a hundred years earlier had been maintained. The question is what is the proper interpretation of these changes that were concurrent with the decline in mortality. From our point of view, they do not support fully the conclusions that Ryder draws and which have been summarized above.

This investigation of the Swedish data illustrates how the study of cohort fertility can be used for analytical as well as for predictive purposes. When the object of research is population forecasts, birth rates are used in order to work with the most current information on fertility trends and to reduce to a minimum the gap between the time of the occurrence of events which serve as a basis for prediction and the time of events which are forecast. Use of these data introduces, of course, the troublesome problem of timing of births that we already have discussed. When the goal, on the other hand, is investigation of the association of selected factors with fertility rates, number of children ever born can be used successfully and for cohorts of completed fertility there is no problem of timing to be taken into consideration. The two types of research are complementary, for results obtained in analytical studies

[8] *Ibid.*, p. 78.

[9] Some of these data have been deduced from the graphs. Not having access to the research materials, the writer is unable to verify these deductions or to make them more precise.

should help forecasters of population trends to judge better the necessity or desirability of a further breakdown of cohorts beyond the age, parity, and marital status subdivision usually used, to discount properly the influence of unusual social and economic conditions upon birth rates, and to estimate correctly some of the long-term trends in family formation and child rearing. Since correct analysis of factors associated with number of children ever born to women of completed fertility depends upon extensive subdivision of the data, the number of cases in the sample must be very large. The only source of such data is usually the decennial census. When comparable data from several censuses are published, trends can be ascertained and often the significance of results can be evaluated properly only with reference to the rate of change in the associations studied. During the past decade, the Census Bureau has published several fertility reports that give in detailed form sample data from 1910, 1940, and 1950 censuses.[10] What, then, are the analyses that have been made or that are currently being made of this large body of raw materials?

A Census monograph on American fertility is being prepared by Kiser, Whelpton, and Grabill using data from these reports.[11] Among tentative chapter titles are: historical setting, ecological aspects, social and economic variations, and other factors in differential fertility. Analyses will be made of the changing size of the American family, rural-urban reproduction, Negro-white differences, and social and economic factors related to fertility. Among the latter will be income, education, and duration of marriage. A feature of these analyses will be use of parity-specific rates for cohorts, carrying forward some of the earlier work done by Whelpton. We have been cautioned, however, by Grabill that the monograph will be a straightforward descriptive study rather than one that explores advanced hypotheses of the kind developed in the Indianapolis Study.

This monograph is a long overdue analysis of the wealth of data from the 1910 and 1940 censuses as well as a timely study of the more recent information reported by the Census Bureau. Very likely, analyses made of the changing size of American families will be more specific and definitive than the study of social and economic differentials in American fertility. Simultaneous cross classification by age of marriage, occupation of husband, education, etc. that would allow further clarification of the interrelation of these variables is not given in the Census reports. We expect, however, that the monograph will be useful as a reference for background information needed in carrying out more theoretical analyses of these factors. We also await with interest to see what else the ingenuity of these scholars may produce for us.

Several studies of differential fertility using data from the several censuses have been made during the past decade and presented in the form of magazine articles, chapters in books, doctoral theses, etc.[12] Through these investigations, we have learned the approximate size of differences in fertility associated with regional and rural-urban residence, with color, and with socio-economic status as indexed by occupation of husband, education of wife, or income. We have seen also how these differences tended to narrow from 1910 to

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[10]U.S. Bureau of the Census: (1) *Sixteenth Census of the United States:1940, Population, Differential Fertility 1940 and 1910,* (a) *Fertility for States and Large Cities,* 1943; (b) *Standardized Fertility Rates and Reproduction Rates,* 1944; (c) *Women by Number of Children Ever Born,* 1945; (d) *Women by Number of Children Under 5 Years Old,* 1945; (e) *Fertility by Duration of Marriage,* 1947; and (2) *United States Census of Population: 1950, Special Reports:* (a) *Fertility,* 1955; (b) *Duration of Current Marital Status,* 1955. All published by the U.S. Government Printing Office, Washington.

[11]Milbank Memorial Fund, *Current Research in Human Fertility,* "Progress Report on Fertility Monograph" by Wilson H. Grabill, pp. 82-92.

[12]See for example, Charles F. Westoff, "Differential Fertility in the United States: 1900 to 1952," *American Sociological Review,* Vol. 19, No. 5, October 1954, pp. 549-561.

1950 first through the higher rate of fertility decline from 1910 to 1940 of the most fertile subgroups and secondly through the greater increase from 1940 to 1950 in the birth rate of the least fertile sections of the population. These studies have been disappointing in several respects: (1) considering the wealth of data, they have been relatively few in number and of limited scope; (2) they have not been successful in isolating the influence of crucial variables by sufficient breakdown of the data or by other techniques; (3) little or no attention has been paid to patterning of the variables and to the collective influence of important factors in constellation form; (4) there has been a general neglect of the influence of age of marriage and of duration of current marital status upon number of children; (5) interrelationships of occupation of husband, education of husband, and education of wife in their influence upon number of children ever born have not been investigated although the distinctions that might be found probably would be useful in further understanding the ways in which socio-economic status and social roles affect fertility; and (6) they have provided only thin interpretations of the existence of fertility differentials and of the changes in them from 1910 to 1950. To some extent, of course, the failures as listed above may be attributed simply to the fact that very little effort has been made to exploit the data.

This last observation might raise the question of whether the census data are sufficiently rich to merit exhaustive analysis. Let us consider first the fertility reports embodying the 1910 and 1940 samples. From an ideal standpoint, we find that the data are inadequate in not giving a simultaneous breakdown of native white women by age, age at marriage, rural-urban residence, marital status, occupation of husband, and education of wife. Without such subdivision of the data, we are unable to isolate the influence of each of these important factors upon number of children ever born. How practicable this breakdown might be is best known to the Census Bureau experts. If necessary some compromise might be effected whereby only cells with a minimum number of cases would be published in detail with remaining cases grouped together in a few residual categories. If the budget of the Census Bureau has been too small to give us the additional breakdown of the data as described above, we would prefer to obtain these data and to dispense with some of the tables that have been published.

Such criticism of the 1910 and 1940 census data is from the point of view of using them to determine the influence of selected factors under specified social and economic conditions upon fertility. It is assumed that the results of such analysis would have theoretical relevance and would be helpful in making population forecasts. This approach, however, is not the only one which can be taken toward the data and which would serve worthwhile purposes as the forthcoming monograph of Kiser, Whelpton, and Grabill will probably demonstrate.

Although our criticism applies with equal force to the 1950 census fertility data, we should be thankful in this case to have any sizable body of published materials since government funds were inadequate and publication had to be financed by private groups. Selection of the limited data, therefore, was probably guided primarily by the research objectives of these groups. These data, nevertheless, will be useful for studies of differential fertility of the kind that has been made during the past generation. Such new studies when made will likely add perspective to our old picture of subgroup differences, but will be unable to unscramble the combined effects upon fertility of the socio-economic variables such as income, occupation of husband, and education of wife.

What research can be undertaken with the 1950 material has been hampered by

lack of strict comparability with data from the earlier censuses. The change in definition of urban residence and lack of funds to present data in all tables for both old and new definitions is an understandable complication. There were other changes in the 1950 tables, however, which have not been explained and for which reasons are not apparent. Consider, for example, some of the difficulties found in an analysis of number of children ever born by education of wife, using Table 9 in the 1940 report on fertility by duration of marriage with Table 21 in the 1950 report on fertility or Table 47 in the 1940 report on women by number of children ever born with Table 20 in the 1950 report. In the first place, the 1950 data are for all whites, whereas, the 1940 data are for native whites. Secondly, educational classes in the 1950 report are dissimilar from those in the 1940 publications with the result that instead of six to eight groups found in one or the other census there are only four or five left after necessary combinations for comparability. And, finally, when analysis by parity of woman is made, the 1950 age breakdown 55 to 59 years is found to have no counterpart in the 1940 report, which gives data for women 55 to 64 years old and 65 to 74 years old. Some of these problems can be solved partially by the researcher, but others cannot be and errors introduced by non-comparability must be estimated.

With refinement of analyses of differential fertility and cohort fertility will probably come further studies of the relation of predicted numbers to resources, to the level of economic activity, to personality development, to family adjustment, and to other matters of concern to society. Interpretation of the meaning of population changes in terms of resources to support increased numbers has been long an interest of demographers and other scientists. One of the recent publications in which the problem is discussed and means for its solution advanced is that by Paul S. Henshaw with the title *Adaptive Human Fertility*.[13] This book is principally of a nontechnical character and has little original evidence to distinguish it. The description of modern methods of fertility control is the best section of the work, as one might well expect since the author's background is mainly that of a biologist. Some students might find informative and interesting his weaving together of factual materials on population and resources with normative considerations in the belief that such presentation makes for a well-rounded understanding of the problem and is best suited for obtaining desirable action.

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[13]Paul S. Henshaw, *Adaptive Human Fertility*, McGraw-Hill Book Co., N. Y., 1955.

THE PROSPECTS FOR GENETIC IMPROVEMENT*

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LOOKING AHEAD IS ONE of those risky occupations which can only be faced by doing just the opposite, namely by looking backward. What we shall know of the future will certainly be determined by what we have learned in the past; and the real problem set for a scientist when you ask him about the future is to extend beyond the present day the lines of progress by which the science of yesterday became the science of today. It requires him to take stock of the significant advances of knowledge in the recent past, and to guess which of these show some prospect of profitable extrapolation in the future.

Of me it is required to take some stock of recent work in the field of genetics, the scientific study of heredity and variation. Heredity, of course, is the biological connection between the generations of plants, animals and men, the bridge over which passes the living substance which we inherit from father and mother and remoter ancestors. This is the substance contained in the egg and sperm from which each of us arose; and it consists of an organized assemblage of thousands of living elements known as genes. The particular system of genes received by each individual—and each individual receives a different set—determines the kinds of responses we make to our environments. The peculiarities of each animal or plant or man are determined by this interplay or interaction between his genes and his surroundings, between his heredity and his environment.

Variations are novelties which arise in one component of this interacting system; they are either due to changes in the genes, known as mutations, so that a new gene-form responds in a different way to the same environment, or to changes in the environment, so that the response of the same set of genes is different. A newly arisen hereditary character like albinism, or spotted skin, or woolly hair, which expresses itself similarly in the usual environments, is an example of mutation; darkening or freckling of skin on exposure to sunlight is an example of a so-called environmental variation, although both kinds of variations, of course, are expressions of a particular system of genes (known collectively as the genotype) reacting with a specified environment or range of environments. The expression, that is, the appearance or behavior of the individual, is known as the phenotype. One of the main purposes of genetics is to understand the relations between genotype and phenotype.

Now we discuss the meaning of another term in the title: improvement. What we shall mean by improvement is something more advantageous for man than what he has now. However much we may speak of knowledge for its own sake or science for the sake of science, the ends of both are certainly and inevitably, although sometimes indirectly, for our human purposes, the betterment of the human condition and the increase of human happiness. The job of the scientist is to search out the rules of order by which nature operates. His domain is the real world of atoms and subatomic particles, their motions and actions whether in molecules, in genes, in living cells of animals or plants or men, in the crust of the earth or in the atmosphere or interstellar space. In this search his im-

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mediate purpose must always be to seek for reason and for order, even though the eventual end of human betterment has to be superseded or even sacrificed in the process. But he must from time to time do what we are asked to do tonight, come out of the laboratory or the study and make some assessment of where we are and where we are going.

Where then are we going genetically? What are the prospects of improvement of the genotypes of the animals and plants upon which we depend both for food and for instruction? (I put instruction almost on a par with food, for we must learn largely from the other living things which we can control experimentally, something we are prevented from doing with our own fellows by moral and practical restrictions.) What finally are the prospects of improvement in human genotypes? Concerning our own conservative, slow-breeding species, I should remind you that man, as man, has a long history of some 600,000 to 700,000 years and during this time no major changes in his structure have occurred, major in the sense that there is no evidence of a tendency for *Homo sapiens* to split into different species. What has occurred, is occurring today and will probably continue, is evolution on a comparatively minor scale zoologically, the formation of races and other more or less distinct groups of genotypes as a result of geographic and cultural isolation, accompanied by separation of populations, inbreeding, and adaptation to different local environments. We should, therefore, not expect any spectacular changes in the human animal in the two generations intervening between now and 2,000 A.D., an interval comprising only about 1/100,000 of man's history. However, changes in the proportions of a few of the many thousands of genes which comprise the total genotype are occurring within and between some human populations today and this is evidence, as I shall show, that such evolution is certainly continuing in our own species.

One way of discussing these questions is to draw up a kind of balance sheet of genetic improvements and disimprovements which we know from experience or by good inference to have occurred or to be occurring now in man or the animals and plants on which he depends. I hope to show you that to discuss either side of this balance sheet apart from the other has not much meaning, for all living things, man certainly not excepted, are parts of nature, which is a continuum of interacting elements in some kind of equilibrium. The balance of nature is no figure of speech. When the frequency of spinsters (in Darwin's old fanciful example) is connected with the quantity of the crop of red clover seeds through steps which lead through cats (kept by spinsters), field mice (hunted by cats), bumble bees (whose nests are destroyed by field mice), red clover blossoms (fertilized by bumble bees), the whole is figurative, but each step (except possibly the first) is a well-established fact. There is, as every ecologist knows, a balance in which predators and prey, parasites and hosts, insects, bacteria, viruses, and food-supply are interacting elements. Disturb one and you disturb the whole chain. So it is with the genotypes of animals, plants and men who all live in populations and societies of which the structure expresses an internal adjustment of the members to each other and of the group to its surroundings.

I shall put down first on the credit side a change in the genotype of human populations in certain parts of the world which is probably occurring today, although the exact measure of it will have to be taken by another generation of geneticists. There is a usually fatal hereditary anemia known as sickle-cell anemia which arises in young children when the child has received from each of his parents a particular form of one gene, known as the sickling gene, since it causes the red blood cells, when under reduced oxygen pressure as in the venous circulation, to assume bizarre crescent-

shaped forms. Such parents have only one such abnormal gene, the other member of the pair being normal, so they do not suffer from anemia, although they can be identified by their blood picture as being carriers of the sickling gene, or having (as medical men say) the sickle-cell trait. This abnormal gene is common in parts of Africa (sometimes 40 per cent of a tribal group will have it) and in Sicily, and in their descendants in the United States, and around Lake Kopais in Greece, and large numbers of children regularly die from this terrible anemia. This gene is most frequent in areas where malicious malaria (*falciparum*) has been hyperendemic; that is, where nearly everyone has been exposed to the malarial parasite. One direct experiment by a British physician in Africa has shown that persons with one of the abnormal genes, (the trait-carriers), are actually more resistant to malaria than related persons with two normal genes. It appears that such carriers are at an advantage in malarial areas, so the frequency of the bad gene increases, and with it the number of children fated to suffer and probably to die from sickle-cell anemia. Malaria, as we know, has been eradicated in America and Europe (including Sicily) by using DDT to kill the mosquitoes which transmit the malarial parasites from person to person. Persons having one sickling gene no longer have an advantage over normal people in Sicily, or in the United States, and we expect the frequency of the gene to fall, and with it the death-rate of children from sickle-cell anemia. This should also happen in Africa when malaria is eradicated there. If it does, we shall have a most dramatic example of an undoubted improvement in a human genotype, obtained as a byproduct of an improvement of the environment by good public health methods. A similar story may be enacted with another hereditary anemia, that is, thalassemia, or Mediterranean anemia, from which thousands of children die each year in the formerly malarial areas of Italy, Sicily, Sardinia and Greece, and in the United States where intermarrying descendants of immigrants from these areas have preserved the bad gene and will for several generations to come. But eventually these hereditary diseases will be rare all over the world.

Let us ask now what flies may get into this apparently good ointment. What if the mosquitoes should become resistant to DDT, as some other insects have done, by survival of those with mutations conferring immunity? There is no sign of this yet, but all of this knowledge is only of recent date, and this is a possible, if not probable, sequel.

There is, however, an additional weight on the improvement side of the scale in these cases, because persons who will transmit either of these genes can be accurately diagnosed by a blood-test. If each such person would refrain from having children, all present anemia genes would disappear from the populations, and we should have to fear only those which arise anew by mutation, probably a rare occurrence.

This suggests, of course, that if we could recognize, *in advance*, those persons who will transmit to their children hemophilia and other diseases known to be caused by an identified gene, such as certain forms of idiocy and imbecility and some other physical abnormalities, the means would be at hand for reducing such genes also to the status of rare genes of which the only examples would be those newly arisen by mutation. There are signs that this may occur for some genes well before 2000 A.D. The improvement will occur only if the carriers of such genes have first the knowledge, second the means, and third the will, to refrain from having children. Alas, this knowledge has been available for certain dominent abnormal genes (expressed when received from only one parent) for a generation, but these genes are still with us. Either the means or the will or both are still lacking. With the spread of the knowledge and practice of contraception,

which is certain to increase, some of this knowledge may be implemented.

These eugenic proposals cannot be efficiently applied by merely preventing the reproduction of persons suffering from hereditary diseases or abnormalities, usually rare, which express themselves only when two like genes (known as recessives) are received, one from each parent. Nature now prevents most such persons from reproduction, but the recessive genes persist in the parents where they cannot be detected until too late, after the afflicted child has been born. However, if persons known to carry such genes would voluntarily refrain from having *as many children* as persons who do not carry these genes, some progress in reducing their frequency might be made.

We shall cite briefly only one other case of improvement in a genotype to show that some probable improvements may be of a temporary character and perhaps balanced by disadvantageous outcomes. The past century has seen a great growth of cities and the consequent break-up of the village communities in which many western peoples lived and married. Certain abnormal genes have been found to occur in a few such small marriage circles or isolates, and when two people, probably related, from the same village marry, there is a greater risk that they should each transmit the same abnormal gene, received from a common ancestor. When such persons marry in the city, there is a better chance that they will marry someone without the same abnormal gene. This reduces the likelihood of the birth of a child with two genes of the same abnormal form; hence the frequency of such an hereditary abnormality should fall. But now the bad genes are more widely distributed and less likely to be reduced by the death of diseased children, each of which carries two of the bad genes out of this world. They thus tend to escape from the control of natural selection which kept them in check, although at a cost of suffering and death, in the villages. But if genes with highly favorable effects under all environments should become more widely dispersed, the urban concentrations of the future, provided they are real melting pots, might confer biological blessings beyond the mechanical, social and esthetic ones by which we profess to justify our choice (generally involuntary) of a metropolis with dirty subways, littered streets and a generally hectic and precarious life.

I have only a little time for two topics which I am certainly expected to discuss, but these I believe involve applications of principles which I have tried to illustrate in the above examples. One, of course, is the effect of the use, both military and industrial, of atomic energy, which is bound to increase the hazards of radiation-injury to the genotype, from which we now suffer to a limited extent due to the so-called normal or background radiation from a mildly radioactive planet and universe, and at the hands of some incautious or X-ray-happy physicians. There seems little doubt that these hazards will increase, especially if some super-bombs go off anywhere above our continent or in our waters. There is no doubt either that increases in the radiation to which living organisms are exposed are followed by increases in the rate of gene mutation, and that most newly arisen mutations have deleterious effects. I do not belong to the prophets of doom, but I cannot view as biologically bright a future in which these hazards will increase. It is true that populations adapt their biological structures in the long run to new hazards and new mutations—but how long is a "long run" for our species? And how much immediate suffering can we tolerate? If we have a choice, none of us would choose to invite the risk implicit in such horrible engines as the H-bomb from which we (and *we* means all human beings) may expect only disimprovement for our species. Meantime we must develop pre-treatments which will counteract the mutagenic effect of radiation on our tissues and germ cells, and ways to do this have already been ex-

plored. Before we descend into the deep shelter, we shall have to take our pills or a nitrogen tank down with us, and it may be that there is no hiding place down there.

For the captive species on which we ourselves are parasites, especially our crop plants, there is some hope, predicated on solid achievements especially of the Swedish plant breeders, of further improvements in yield and quality through the use of carefully controlled radiation. Already grain-varieties have been produced by selection and recombination among radiation-induced mutations which extend the range of profitable crop production several degrees northward, and there is no reason why this method should not yield further improvements. But I suspect that the mutation process involves chemical steps which will provide more effective and less risky entreés to the great problem of the future in this field—the deliberate control and direction of mutation in specific genes or groups of genes. We shall certainly be nearer to that goal by 1975. Perhaps it will be achieved by 2000.

The second point of these two is: what are the prospects for changes in the genotypes of the world of microscopic life on which we depend and which holds over us continual threat and hope—the world of protozoa, bacteria and viruses? Here the fact of balance is clearly established. No sooner had we gained control over some pathogenic micro-organisms through the use of antibiotics than we found the parasites able quickly to adjust themselves to the new hazard. Micro-organisms have survived so many such hazards in the past, that they have evolved as a result of their history a plasticity due to random mutation by which, if resistance to streptomycin is advantageous, lo, the mutations are already present, and such mutant types survive as streptomycin-resistant strains. More than that, some of them now require streptomycin and cannot survive in competition without it. We keep one jump ahead only by discovering new antibiotics, by inducing mutations in the molds that produce the antibiotics (such as penicillin) so that they produce a greater quantity of antibiotic, and by learning the metabolic systems of the micro-organisms with which we must continually contend. I believe we are smarter than they and shall win. But we may kill some good friends in the process, for many of the constructive activities of plants, animals and men cannot go forward without some of these symbiotic beneficent allies. Meantime we have, by applying the same principles, conquered some of the most formidable bacterial and virus diseases of crop plants by breeding genotypically resistant varieties, one of the great triumphs of plant-breeding. The field of the genetics of micro-organisms is one of the currently brightest spots of the whole science, chiefly because new ideas about the gene itself are coming from it, and this glow falls especially on the future, for where new ideas are now, new facts will follow, and after that a new order which brings us nearer to our goal.

This brings me to my last point, and for this I should like to abandon the checks and balances of the ledger and speculate with more freedom, though I hope not with less responsibility. What does the biologist whose tools are genes speculate about when he leaves the laboratory and stands before an audience like this? What are his reasoned hopes for the next 50 years? I have already made some optimistic statements about prospects for controlling the genotypes of the most rapidly reproducing organisms, bacteria and viruses, and as a consequence, reducing the toll of disease on human and especially crop-plant populations. Two useful organisms with annual generations—Indian corn and chickens—have been greatly improved in the past 25 years, both by selective breeding and by controlled heterosis, the bonus of vigor and yield obtained in first generation crosses between different genotypes. This can certainly be extended to other cross-breeding plants and animals. New sources

of food will probably be made available by favorable mutations in unicellular green algae yielding cheaper carbohydrate, and in yeasts, yielding more and better proteins. Now that we know that the chief organic catalysts, the enzymes, are controlled by genes, we can surely harness these for human purposes.

The greatest recent increase in our knowledge of human genotypes has come from the combined efforts of serologists and geneticists working on human blood group genes, of which a dozen major systems are known, some of which consist of many elements, so that by combination of these literally millions of different genotypes become possible. The day is not distant when the uniqueness of each human individual will be proved from his blood group genes alone. As a byproduct of this knowledge one cause of infant mortality, hemolytic disease of the newborn, is now controllable. Relations between some of the other blood group genes and specific diseases (for example between blood group O and duodenal ulcer) have recently been proved. Others are certain to follow quickly. Genetics has entered medicine, and diagnosis of human genotypes must soon become an essential part of the practical knowledge of the physician. Human genetics will shortly have an efflorescence which will put it in the forefront of the sciences contributing to human welfare, with profound social effects.

I shall not try to summarize this review and preview except to point out that wherever improvements, past or prospective, appear, they are associated with (or I suppose judged by) increased control and increased efficiency or effectiveness of scientific method, and this constitutes empirical knowledge, if not understanding. On the other hand, the other side of the ledger shows, on balance, the greater precariousness which is to be, for better or for worse, the condition which will dominate the existence of our descendants in 2000 A.D. That is a condition which ignorance, unreason, prejudice, even lack of rapid scientific advance, could quickly make entirely untenable for civilized man. Once we have got the jump on nature, we shall have to keep on jumping faster and farther. This is to say that we shall become increasingly more dependent on our wits, those specifically human attributes such as tool-using, and the use of language with its corollary, thought—in short on cultural development. Surely that is no novel conclusion. Twenty-five hundred years ago an anonymous preacher told his people: "Wisdom is the principal thing; therefore get wisdom; and with all the getting, get understanding." Today we should remember that more of that task lies ahead of us than behind us.

HEREDITARY SUSCEPTIBILITY IN RHEUMATIC FEVER

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RHEUMATIC FEVER IS AN IMPORTANT, relatively common disease having its usual onset in childhood. The heart is usually affected. (1) It has been estimated that for each death of a person under 20 years of age due to the combined causes of pertussis, diphtheria and poliomyelitis, there were 50 deaths due to rheumatic heart disease (2).

The etiology of rheumatic fever is obscure, and its incidence in the population is not obtainable. As it is a disease which appears to "run in families," the family unit has offered the best approach for genetic studies.

It is an accepted fact that certain individuals are more susceptible than others to the acquisition of many diseases. In recent years it has become clear that the character of the host is one important factor in resistance and susceptibility. The biological pattern of development of an individual is determined both by his hereditary constitution and his environment during his life experience.

For more than fifty years the clinical impression that heredity is a significant factor in the concentration of rheumatic fever in certain families has been widespread. This belief was based in large measure on the observed familial incidence of the disease (3).

There have been several family studies in rheumatic fever. These have considered the relative roles of environment, contagion, and heredity as factors responsible for the high familial incidence of the disease. More recent family studies have demonstrated a higher degree of familial association in families with parental rheumatism, and a greater frequency of cases among the progeny of rheumatic families than controls.

Modern Genetic Theory

The study of human heredity as a medical subject is a relatively new field. In the past there have been a number of purely genetic studies on rare conditions. In recent years, medical genetics has assumed a prominent place in clinical investigations. The application of genetic techniques to human material involves certain difficulties but is not, for the most part, beyond clinical medicine. The reasoning is simple and the arithmetical details are easy to follow and apply.

Many conditions appear to "run in families," and the observed familial incidence cannot be considered as conclusive evidence for heredity. The attempt to link heredity with familial incidence has been associated with the concept of constitution which embraces a variety of poorly defined traits. Modern genetic theory refers to explicit cellular and functional attributes and properties whose precursors have a concrete and real existence in the germ cells and the soma.

Fundamentally, all statements about heredity refer to the chromosomes upon which the genes are located. Variations in the mechanism of heredity depend on processes associated with the maturation of the germ cells and the fertilized egg. Inasmuch as all hereditary characters probably

depend upon the effect of a variety of exogenous factors for their somatic expression, hereditary traits may be variable in the time and degree of their expression. Not infrequently they may fail to be expressed altogether, even though the host may be genetically susceptible. It is therefore essential to evaluate the effect of nongenetic factors, environmental, dietary, or bacterial.

In the study of any hereditary condition, the pattern of inheritance is described in terms of some one of the known mechanisms of inheritance in man (4, 5). By the use of appropriate formulas which have been derived on the basis of experimental studies, the theoretical distribution of susceptible individuals in a series of human families is made and compared with the actual observation for each family size. If agreement is obtained, the data are considered to be consistent with the hypothesis postulated. Consistency with a postulated mechanism is not in itself proof. Other mechanisms of heredity must be excluded. Significant differences between observation and expectation negate the mechanism postulated. Consistent deficiencies or excesses of cases may indicate nongenetic effects if bias in the data is excluded. For these reasons the requirements of the data suitable for application of genetic formulas are one of the most important aspects of the genetic analysis. The bias introduced and the errors of interpretation arising from a lack of recognition of these requirements in rheumatic fever data must be taken into account.

Methods of Genetic Analysis

The incidence of a disease or condition among twins is frequently used to establish the possible role of genetic factors. It would be expected that traits in which heredity plays a prominent part would not occur so frequently in both members of fraternal twins as in a pair of identical twins. Such studies have given results consistent with the hypothesis that hereditary factors are important in rheumatic fever (6).

Various methods of genetic analysis may be utilized. A simple measure of familial association is a comparison of the ratios of affected siblings to total number of siblings for several matings. Pedigrees or family trees are frequently used to chart the descent of disease or defect in human families. This method has limitations. Estimates of affected progeny for certain common matings is presented in Table 1.

TABLE 1

ESTIMATE OF AFFECTED PROGENY FOR CERTAIN MATINGS

Hereditary Mechanism	Both Parents Negative	One Parent Positive	Both Parents Positive
Dominant	None	*One-half*	All
Recessive	*One-quarter**	One-half	All
Sex-linked	*Sons: One-half* *Daughters: None*	Father + : None Mother + : All sons, no daughters	All

*The mating type (*italicized*) is the one most commonly encountered clinically for each mechanism of inheritance. Negative parents are considered carriers.

In using simple genetic ratios the assumption is made that the clinical data are representative of the matings for each type in the general population. This is rarely attained in ordinary clinical material due to bias in the selection of families for study, for in view of the small size of human families in comparison with experimental material, certain families without any positive siblings should occur in the general population by chance alone. Such families would not be represented in samples drawn from a clinic population which includes at least one case in a family.

The Lenz-Hogben Formula. The use of simple Mendelian ratios for genetic analysis may be expressed in terms of varying family size and total affected siblings by the following formula:

$$s = c$$

$$R = \Sigma\, Sn_s p \pm \sqrt{n_s k_s}$$

$$s = 1\ 1{-}q^s$$

R = Total cases expected
c = Maximum family size
s = Family size
p = Frequency of affected offspring, *i.e.*, the genetic ratio
q = Frequency of normal offspring, when $q+p = 1$
n_s = Number of families of s size.
Sn_s = Total siblings in families of s size.

$$k = \frac{s\,(1-q)}{(1-q^s)^2} \times [sq^s(q-1) + q\,(1-q^s)]$$

The application of the Lenz-Hogben formula for predicting the total number of cases in a series of families is simplified by the use of genetic factors (7), as shown in Table 2.

TABLE 2

GENETIC FACTORS FROM THE LENZ-HOGBEN FORMULA FOR USE IN GENETIC PREDICTION*

Family Size	Both Parents Negative (q .75, p .25) $sp/1{-}q^s$	K_s	One Parent Positive (q p .50) $sp/1{-}q^s$	K_s
1	1.000	0.000	1.000	0.000
2	1.143	0.122	1.333	0.222
3	1.297	0.263	1.714	0.490
4	1.463	0.420	2.133	0.782
5	1.639	0.592	2.581	1.082
6	1.825	0.776	3.048	1.379
7	2.020	0.970	3.528	1.667
8	2.222	1.172	4.016	1.945
9	2.433	1.380	4.509	2.215
10	2.649	1.592	5.005	2.478

*Values of k_s are computed from Hogben's formula.

Values of $s/1{-}q^s$ have been computed from Lenz-Hogben formula for the several values of p and q in our data; ($sp/1{-}q^s$ may be computed for each family size and applied as a genetic factor for prediction).

To compute the expected number of cases in a group of rheumatic families of given size, the appropriate factor is multiplied by the number of families of that size (n_s). The standard error (σ^2) is obtained by multiplying the appropriate value of k_s by the number of families. For example, the expected number of cases in the two-membered families in our data is obtained by multiplying the factor 1.143 by 9, the number of two-membered families. Its variance (σ^2) is obtained by multiplying 9 by 0.122. This procedure is followed for every group of families arranged according to family size and parental diagnosis.

Probably the best proof of the hereditary mechanism would be available from an analysis of families selected because one or both parents were rheumatic. These data are preferable for genetic analysis since they are not biased by selection of affected children and can therefore be analysed directly by the application of simple Mendelian ratios.

Genetic Studies in Rheumatic Fever

Genetic and epidemiologic studies in 1937 and 1943 (8, 9) revealed that the distribution of cases among 471 children in 113 rheumatic families was consistent with a genetic mechanism of simple recessive inheritance. These families were selected because of the presence of at least one rheumatic child. It was concluded that hereditary susceptibility is the primary factor responsible for the concentration of rheumatic fever in certain families.

An hereditary factor in rheumatic fever has been reported by other investigators. In 1952, Gray, Quinn and Quinn reported agreement with a simple recessive gene hypothesis in 40 New Haven families similarly selected (10). There was a paucity of cases in three positive by positive matings. In a Toronto study by Uchida in 1953, it was concluded from a genetic analysis of 58 affected families that no definite mode of inheritance could be established from the limited data (11). In 1953 Stevenson and Cheeseman studied 462 families in Belfast which were ascertained by an affected child, and 51 families ascertained by an affected mother (12). It was concluded that inheritance played a major role in determining familial aggregation of cases but that a Mendelian mechanism could not be established.

Further investigation of the pattern of inheritance in rheumatic fever was under-

taken by Wilson and Schweitzer in 1954 (13). The families of a selected parent were brought under observation without regard for the presence or absence of rheumatic fever in the unselected mate or in collateral relatives.

Pattern of hereditary susceptibility in rheumatic fever. The genetic analysis of 109 rheumatic families in 1943 is presented in Table 3. Among fifty families in which one parent was rheumatic ($p = 0.50$), the observed frequency of 117 rheumatic children is not significantly different from the expected value of 111.9 ± 6.43, the deviation being $0.79\ \sigma$. In the families with two non-rheumatic parents, 101 cases among 251 offspring were observed in comparison with 90.0 ± 5.35 cases expected ($p = 0.25$). The deviation is on the borderline of significance, $2.06\ \sigma$.

The use of the Lenz-Hogben formula to estimate the number of cases which should be observed is illustrated by this analysis. The agreement obtained, assuming theoretic frequencies of 0.25 and 0.50, constitutes presumptive evidence in favor of a simple recessive gene mechanism in rheumatic fever.

There was no agreement when mechanisms such as a single dominant gene, sex-linkage, two or more recessive factors, or more than two dominant factors were postulated. Although the operation of a double dominant gene was not entirely excluded, application of this hypothesis to the data gave an estimated frequency of 8 per cent for the disease in the general population, a value far higher than is estimated by current observers.

The assumption that the mechanism is a single autosomal recessive gene is consistent with the data and with the general observed incidence of the disease. It is also the simplest concept which may be postulated. It was therefore felt that for purposes of further analysis, this genetic mechanism should be assumed and, if correct, the results would substantiate it.

In our 1954 study the following analysis was made. Families of a selected parent were brought under observation without regard for the presence or absence of rheumatic fever in the unselected mate or in collateral relatives. The data were arranged

TABLE 3
CASES EXPECTED AND OBSERVED USING THE LENZ-HOGBEN FORMULA

Family Size	Number of Families	Total Children	Rheumatic Children		Difference	σ^2	Difference/σ
			Observed	Expected			
			A. Both Parents Negative				
1	1	1	1	1.0	0.0	0.00	0.00
2	9	18	11	10.3	0.7	1.10	0.67
3	14	42	20	18.2	1.8	3.68	0.94
4	12	48	19	17.6	1.4	5.04	0.62
5	10	50	20	16.4	3.6	5.92	1.48
6	5	30	12	9.1	2.9	3.88	1.47
7	3	21	9	6.1	2.9	2.91	1.70
8	4	32	8	8.9	—0.9	4.69	0.41
9	1	9	1	2.4	—1.4	1.38	1.19
Total	59	251	101	90.0	11.0	28.60	2.06
			B. One Parent Positive				
1	6	6	6	6.0	0.0	0.00	0.00
2	7	14	10	9.3	0.7	1.55	0.56
3	11	33	21	18.9	2.1	5.39	0.90
4	9	36	22	19.2	2.8	7.04	1.01
5	3	15	9	7.7	1.3	3.25	0.72
6	6	36	17	18.3	—1.3	8.27	0.45
7	2	14	7	7.0	0.0	3.33	0.00
8	3	24	9	12.0	—3.0	5.84	1.24
9	3	27	16	13.5	2.5	6.64	0.97
Total	50	205	117	111.9	5.1	41.31	0.79

in parental mating types and appropriate Mendelian ratios applied. Standards tests for dominant, sex-linked, and recessive inheritance were done.

Genetic analysis supported the single recessive hypothesis and excluded sex linkage, dominant and multiple-gene hypotheses. A comparison of the number of cases so far observed and the number expected for different parental family types is presented in Table 4. The number expected at the present age was obtained by application of an age correction factor. There were 52 families which were considered genetically susceptible and in this group there were 40 cases observed among 121 siblings. The familial incidence was negligible in the genetically nonsusceptible families. Only 3 cases were observed among 525 children in 239 families.

It will be noted that of 49 cases finally expected, 40 have so far been observed. Since all of the siblings had not passed the peak age of onset of the disease, the data were corrected for present age. On this basis, 45 cases are expected at the attained age and 43 are observed. Good agreement is noted at the attained age of the children. A few additional cases are expected in the susceptible families as the younger children reach the ages of maximum incidence. It is unlikely that any considerable number of new cases will occur at older ages. In the earlier study, 130 individuals who were nonrheumatic at the ages of 13 to 25 years have remained non-rheumatic during 11 additional years.

TABLE 4

COMPARISON OF THE NUMBER OF RHEUMATICS OBSERVED AND EXPECTED BASED ON TEST FOR RECESSIVE INHERITANCE.

Mating Type	Families	No. of Children Siblings	Under 6 Years	Cases Observed	Cases Expected
(a) $+ \times +$	8	15	2	14	15
(b) $+ \times C$	7	20	0	10	10
(c) $+ \times C^{\bullet}$	28	63	8	11	21
(d) $C \times C$	2	4	0	1	1
(e) $C^{\bullet} \times C^{\bullet}$	7	19	4	4	2
	52	121	14	40	49
(f) $+ \times -$	183	399	80	2	0
(g) $C \times -$	30	65	13	0	0
(h) $- \times -$	26	61	12	1	0
	239	525	105	3	0
	291	646	119	43	49

(a) Positive mated with positive; in this mating all children are expected to be positive.
(b) Positive mated with negative who is the child of a rheumatic; half of the children are expected to be rheumatic.
(c) Positive mated with negative who is the sibling of a rheumatic; one-third of the children are expected to be rheumatic.
(d) Two negative individuals who are both children of rheumatics; one-quarter of the children are expected to be rheumatic.
(e) Two negative individuals, both of whom have some close kin who are rheumatic; fewer than one-quarter of the children are expected to be rheumatic.
(f) Positive mated with negative who has no rheumatic parentage or close kin (siblings, aunts, uncles, nieces and nephews); no rheumatic children are expected.
(g) Negative who has a rheumatic parent or sibling mated with a negative with no rheumatic parentage or close kin; no rheumatic children are expected.
(h) Both parents negative, with no rheumatic parentage or close kin; no rheumatic children are expected.

$C^{\bullet}$—nonrheumatic parent who had a rheumatic close relative other than parent. C is a nonrheumatic parent who had one rheumatic parent.

Comment

Genetic analysis of human data is limited by the relatively small family size as compared with experimental material. This may preclude complete expression of an hereditary trait. In addition, diagnostic difficulties may prevent adequate ascertainment of cases. This is particularly true in the diagnosis of rheumatic fever in adults since physical signs of rheumatic heart disease often regress and childhood illness is not accurately recalled. In the absence of a specific diagnostic test, complete reliance is frequently placed on historical information or a limited physical examination. In this study, evidence of rheumatic heart disease determined by physical, fluoroscopic and electrocardiographic examinations was required for confirmation of a diagnosis of rheumatic fever. In spite of the rigid diagnostic criteria utilized, there were probably a few errors in parental classification in

families where information was limited concerning collateral relatives. The continuous medical supervision of the families, the majority from infancy to past the peak age of onset, minimized possible diagnostic errors in the children.

The conclusion that the pattern of inherited susceptibility is a recessive character rests on the agreement between observation and expectation. Equally important for the interpretation of recessiveness are the observations in the lower half of Table 4 (f-h). The insignificant number of cases in 239 families where one parent is non-rheumatic and not known to have close kin who are rheumatic is strong evidence that recessive inheritance is involved.

Complete agreement with a specific hereditary mechanism may not always be obtained even in conditions of proven heredity. It is well known in experimental material that certain modifying factors, endogenous or exogenous, may influence the expression of hereditary traits. This may also occur in human disease. The differences in type and severity of manifestations of rheumatic fever among members of a family suggest differing degrees of susceptibility which perhaps reflect endogenous modifying influences. There are probably also exogenous factors which may influence the expression of the disease in a susceptible child.

The adequacy of recessive inheritance to describe the distribution of cases among these families does not exclude a more complicated mechanism. It is, however, the simplest hypothesis, requiring no additional assumptions.

The results of these investigations in two family generations support the concept of single recessive inheritance. It may be noted that the analytic procedures in the two studies were different. The procedures in the second study are more satisfactory because they do not require the statistical adjustments that must be made when the affected child is the index case. When data are collected based only on parental selection, the standard Mendelian methods of analysis are fully adequate. Where information on the antecedents of the parents is meager, statistical analysis based on estimates of population frequency is used to determine the pattern of inheritance. This method of analysis requires a series of families collected as a random sample. This important condition is not fulfilled in clinic families.

The primary role of heredity in the familial concentration of rheumatic fever has been established in this and other published investigations. These observations emphasize the importance of identifying the genetically susceptible family to facilitate early diagnosis and treatment. The medical supervision of these families affords an opportunity for investigation of the nature of the inherent factor responsible for susceptibility. They are also useful for the study of exogenous factors which may be responsible for the development of rheumatic fever in a susceptible child. Current etiologic concepts may be explored within this framework.

The demonstration of hereditary susceptibility does not exclude the operation of environmental factors. Like most published family studies, our investigations concern a clinic population which excludes families of higher economic level. The economic status and living conditions which prevailed in 1955 among the susceptible and non-susceptible families was comparable and far superior to those in the earlier study in 1943.

These studies indicate that in the general population there is a distribution of children who are susceptible or unsusceptible to rheumatic fever on a genetic basis. Genetic analysis gave good agreement with single recessive inheritance while excluding other mechanisms.

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HEREDITY COUNSELING

The Darker Side

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In this article I do not want to give the impression that I am against genetic counseling. I am, of course, very much for it, and enthusiastic about the many ways it may help people. But it needs to be pointed out that there are many pitfalls to trap the unwary, many important factors that the inexperienced counselor (including myself) may fail to consider, and many cases where it seems impossible to supply any satisfactory solution. To draw attention to such problems, so that more effort may be devoted to solving them, this paper will discuss some of the unsatisfactory aspects of counseling.

A. Genetic Heterogeneity of Clinical Entities

A lot of difficulty comes from the fact that for many diseases two clinically similar cases may be genetically different, and thus have different genetic prognoses.

Example 1. The patient, aged 13 years, has diabetes mellitus. Three older and four younger siblings are well. Her mother is diabetic, and has one brother and three sisters who are well, and a sister who is diabetic. The mother's mother is also diabetic. Is this a case of "ordinary" diabetes, in which the risk for a subsequent sibling is something over 10 per cent (Steinberg, 1955), or is it a clinically similar disease due to a rare dominant gene, where the risk is 50 per cent? At present there seems no way of telling.

A similar problem may arise in the case of consanguineous matings giving rise to offspring with relatively common diseases.

Example 2. The patient has a cleft lip and palate, and it is found that the parents are first cousins. The consanguinity may be a coincidence, and this case may be no different, genetically, than most other cases of cleft lip and palate, in which the risk for the next child has been estimated as about 5 per cent (Fogh-Andersen, 1942). But we know that not all cases of harelip and cleft palate are etiologically the same (Van der Woude, 1954). The parental consanguinity in the present case suggests that here a rare recessive gene is the cause of the patient's defect, and that the risk for future children is 25 per cent. How can we distinguish between the two possibilities for this particular case— coincidental consanguinity or rare recessive?

For what it's worth, the frequency of cleft lip in the siblings of children with cleft lip (± cleft palate) does *not* seem to be elevated in the rather small sample available. I have selected from three series of cases of harelip and cleft palate (Fogh-Andersen, 1942, Fraser, unpublished, Sanders, 1933) the families in which the proband's parents were related. There were 20 such families, with 66 siblings of the probands, and only 2 (3 per cent) of these were affected. This is not significantly different from the risk for a sibling of a proband whose parents are unrelated (5 per cent).

Parental consanguinity may also raise counseling problems when the patient's condition is rare, but usually not inherited.

Example 3. The patient is diagnosed as having retrolental fibroplasia, a condition

occurring in premature infants, probably due to improper oxygen administration, and leading to blindness. The fact that the parents are first cousins leads one to wonder if, in this case, the eye changes may be due to a recessively inherited disease producing the clinical picture of retrolental fibroplasia. This makes it hard to give the parents any useful advice. If it is the usual type of retrolental fibroplasia the risk of recurrence is very low, but if a simple recessive gene pair is involved the risk is 25 per cent. One hesitates to warn the parents of a rather high risk of recurrence without any real evidence; even to mention, however cautiously, the possibility of a 25 per cent risk of recurrence, is likely to cause them a great deal of harmful, and perhaps unnecessary, worry. But to ignore the possibility of recurrence may expose them to a severe shock if the condition does recur and they have not been warned. Even so, the latter may be the better course, though it does stick the counselor's neck out a bit.

A third reason why genetic heterogeneity in clinical entities causes difficulties in counseling is the existence of phenocopies, where an environmental agent may produce a disease clinically indistinguishable from one produced by a pathological gene.

Example 4. Mr. Dillon* had bilateral retinoblastomas removed surgically when he was 2 years old. This malignant disease of the retina is known to show dominant inheritance in many cases; the "sporadic" cases are thought to be either mutations or phenocopies. Mr. Dillon, whose family history is free of this disease, married a woman who was accidentally blinded as a child, and they have a son who at the age of 6 years has no signs of retinal disease. They were referred for counseling, because they very much want another baby. They are intelligent, loving parents, both college graduates, who are doing a fine job of raising the present child. They cannot adopt a child, as blind couples are not considered suitable adoptive parents by the adoption agency. What shall they be told?

If Mr. Dillon's neoplasm was gene-determined, the baby's chance of losing at least one eye, and perhaps its life, is 50 per cent. If it was a phenocopy, the chances are virtually 0 per cent. Opinions as to the proportion of retinoblastomas that are phenocopies range from almost none to three out of four (Nachtsheim, 1955). Suppose we guess it as one out of four. Then the risk for Mrs. Dillon's future child is 3 chances in 8 (37.5 per cent) of getting a retinoblastoma. This is the statistical probability, but it is not very helpful to the parents. They would, I think, rather be told that "The chances are either 50 per cent or 0 per cent although we do not know which" than that "The chances are 37.5 per cent." But it would be infinitely more helpful if someone would discover how to distinguish the phenocopies from the genetic cases.

Since the parents were intelligent and responsible, they were told more or less what has been written here, and the matter was discussed at some length and from many angles. After much soul-searching they decided to take the risk and Mrs. Dillon is now pregnant. Since the gene usually does not manifest itself at birth, the outcome of their decision will not be clear for several years.

B. Clinical Heterogeneity of Genetic Entities.

The fact that a single gene may have widely different manifestations in different persons, or perhaps no manifestation at all, leads to difficulties in counseling simply because it may be impossible to make a definite diagnosis.

Example 5. A French-Canadian couple was referred by Social Service for an opinion as to the outlook for future children. The first baby, a boy, died at the age of two days, and the parents were told that "the hole in his heart did not close." He was not in a hospital and there was no autopsy. The second child, a boy, died at the age of 5 weeks, of gastroenteritis. He had been "blue" all his

*All names are, of course, fictitious.

life. The record from another hospital stated that he had "circulatory trouble." The third child, a boy, age 7, is well. The fourth, also a boy, had cyanotic congenital heart disease which at autopsy was found to be a most complicated defect involving, among other things, transposition of the aorta and an aplastic left ventricle. The baby had a decidedly dolicocephalic skull, a right inguinal hernia and left hydrocoele. There was no parental consanguinity, and no history of similar heart defects in the relatives.

What then is the outlook for the next child? Three cases of congenital heart disease in one sibship is certainly unusual. Is this a striking coincidence (in which case the outlook for the next child may well be good) or is there some special factor predisposing to developmental heart defects in this family? One thinks of arachnodactyly (Marfan's syndrome), for instance, where an irregularly dominant gene may or may not produce cardiovascular disease along with one or more of a variety of defects including long fingers and toes, an elongated habitus, dolicocephaly, and eye defects such as dislocation of the lens and myopia (Lutman and Neel, 1949). The mother is short and fat, and nothing unusual is found on physical examination. The father is not tall (5'8") but is on the thin side, has a dolicocephalic skull, a long narrow chest, and has a "bad eye." Ophthalmological examination shows there is no dislocation of either lens, but a severe myopia of the left eye. He does not have long fingers, but it has been pointed out that the skeletal effects of the gene may be largely overshadowed in a person who is constitutionally disposed to be short and stocky (McKusick, 1955). If the defects in the children are due to the arachnodactyly gene, the outlook for future children is relatively poor. But we do not know for sure whether the arachnodactyly gene is involved here or not. Certainly it would be hard to convince the average clinician, or a court, that the father represented a form (however *fruste*) of Marfan's syndrome. To complicate matters further, the mother tried to abort this, and previous pregnancies, and feels that the defect is either caused by, or a punishment for, her action. She is very depressed, guilty and hostile, particularly to her husband. What will be the effects on the marital relationship if she is told it's not her fault, since her husband transmitted the gene. (Of course the fact that the husband may have transmitted the gene to the child does not mean that the child's disease is his fault, but that is the way the mother will interpret it.)

The parents were told, during a great deal of discussion, that the defects were certainly not due to the abortion attempts, but that there was a distinct possibility that it might occur again (there was no point in estimating a probability, which might give the impression that we actually knew what the probability was). As they are Roman Catholics, their choice of ways to meet this problem is decidedly limited. I feel that counseling in this case was a failure, partly because we could not give a reasonably definite prediction (since we do not know enough to identify minor forms of Marfan's syndrome with certainty) and partly because what the mother really needed was extensive psychiatric treatment, which we could not supply, and which she was unwilling to request from the proper sources.

So far in this discussion the source of our difficulties has been mainly a result of inadequate genetic knowledge. Many other examples could be cited—for instance, the problem of a hemophiliac with a negative family history and whether or not his mother is a carrier. Or the cases that just do not fit genetic theory, such as a family in which pseudohypertrophic muscular dystrophy shows regular sex-linked recessive inheritance except for the presence of a pair of monozygotic, female twins, of which one member has the disease!

C. Psychological and Emotional Complications.

Now let us consider some of the counseling problems arising not from inadequate genetic knowledge but from the psychological and emotional conflicts that are often involved in counseling situations, and which, if unperceived or ignored by the counselor, may make the counseling procedure worse than useless.

1. Ulterior motives of those seeking counseling.

Sometimes a person who comes for genetic counseling does so not because he is sincerely interested in making a decision on the best available evidence, but because he hopes to use the genetic situation to further his own ends.

Example 6.† A 23-year-old albino French-Canadian man was referred by the Institute for the Blind because he was considering marriage and wanted to know the risk of his children being albino. Two of his nine siblings were also albinos. It was explained to him that the statistical risk for his children was low, but that the crucial point was whether or not the prospective wife was a carrier, the risk being 50 per cent if she was, but 0 per cent if she was not. In view of Waardenburg's claim that albino "carriers" may often be identified by transillumination of the iris (cited by Falls, 1953) we suggested that the fiancée should come in for examination. The patient reacted so strongly against this that further delving into his feelings about the fiancée was undertaken, and it was found that she was eight years older than he, that she, not he, had done the proposing, and that, in short, he was not at all keen on getting married and was hoping to use the poor outlook (as he thought) for the children as a reason for not going into the marriage.

Example 7. Mr. Dixon consulted me because his son, Alan, was intent on marrying Mr. Dixon's sister's daughter. He did not seem opposed to this, but wanted to know if there was anything to the idea that first cousins shouldn't marry. The genetic disadvantage of consanguineous matings was explained, and it was pointed out to him that although marrying a first cousin certainly increases one's chance of having defective children, we really do not know what the actual risk is—if we had to guess it we might say about a 10 per cent risk for each child of developing a recessively inherited disease. This risk constitutes an argument against the marriage which would have to be balanced against the arguments for the prospective marriage, and the decision taken accordingly. Shortly afterwards Mrs. Andrews, Mr. Dixon's sister, consulted me, not knowing of Mr. Dixon's visit, because her daughter was thinking of marrying Alan Dixon. Exactly the same advice was given to her.

The result was that Mr. Dixon went away saying "The geneticist says the risk is low, and there is no objection to the marriage," while Mrs. Andrews went away saying "The geneticist says the risk is high, and the marriage would be ill-advised." Both interpreted the information to suit their own ends.

Because of the possibility of being misinterpreted, it is wise for the counselor to record his advice in writing, although the counseling should, of course, always be done by personal interview.

2. Psychological pressure on the counselor.

At times the counselor may be subjected to pressure from various sources that may tend to influence his decision. It is, in some circumstances, almost impossible to avoid becoming emotionally involved in the situation.

Example 8. Mr. and Mrs. Johnstone were referred by the obstetrician for an opinion regarding the outlook for future children. Their first baby, a girl, had died at the age of 4 days with a tracheo-esophageal fistula. The next child, a girl, had died in hospital with a rare developmental defect of the cardiovascular system. The third baby, a boy, died with

†I am indebted to Dr. J. D. Metrakos for supplying the details of this case.

the same sort of cardiovascular anomaly. The parents were, naturally, quite convinced that they were unable to have normal children, and intended to adopt a family. However, Mrs. Johnstone became pregnant, and when they arrived for counseling they were determined to have the pregnancy terminated. It became clear that the counselor was expected to provide scientific justification for this procedure by stating that the next baby was almost certain to be malformed. However, our evaluation of the situation did not add up this way. There is no evidence of a relationship between tracheo-esophageal fistula and developmental cardiac defects, and no good evidence that the risk of recurrence of congenital heart disease is higher after a mother has had two affected children than after she has had one. We therefore concluded that the risk for the next baby was relatively low. (Note that not all counseling problems involve having to tell parents of the ominous outlook for future children—here it was quite the other way around!) The following is an extract from our letter to the referring obstetrician.

"I am therefore forced to conclude that the disastrous sequence of malformed children in this family should probably be regarded as an unfortunate coincidence. Even if it were due to a hitherto, unrecognized recessive gene the chance that the next child would be affected would be 25 per cent—*i.e.*, the odds would be in favor of getting a good baby. It therefore seems that, unless this represents a previously unrecognized medical phenomenon, the chances are in favor of the next baby being healthy.

"I feel I ought to mention, however, that Mr. and Mrs. — were quite skeptical when I told them this, and quite understandably so. They said, in fact, 'That's what they told us the last two times,' and I could understand how, in spite of the statistics, they might feel convinced that the next baby would inevitably be malformed. They seemed to feel, when I saw them, that Mrs. — might not be able to stand the fear, worry and heartache of the gestational and neonatal period, and that if the child were malformed (it has at least a 3 per cent chance of being defective in some way) the effect on the parents would be nothing short of disastrous."

This decision was reached in the face of an almost overpowering intuitive feeling on the counselor's part that Mrs. Johnstone was a woman who for some reason seemed destined to have nothing but malformed babies. In addition there was the tremendous emotional pressure brought to bear by the parents on the counselor to give a poor prognosis for the next baby. All sorts of appeals to the counselor's sympathy were used, and even the argument that if Mrs. Johnstone had a nervous breakdown under the strain it would be the counselor's fault. It was a harrowing experience all round, and there were several people besides the parents who worried throughout the remaining seven months of the pregnancy. Happily, the baby turned out to be a nice healthy little girl.

D. Other Problems

The problems that the counselor meets in his practice are endless in their variety and present a continual challenge to his sensitivity, tact and ingenuity. Each situation is different and must be approached with the knowledge that the problem is not only a genetic one, but involves domestic, economic, psychological and emotional factors, recognition of which may be vital to the successful management of the case. A few miscellaneous examples will provide further grist to the mill of my thesis.

Example 9. Mr. and Mrs. Laflamme were referred because their baby boy had been admitted to the hospital with amyotonia congenita. The parents were unrelated. Their first child was a girl, now three years old, who is well. The second child died in another hospital at the age of 2 months and was diagnosed as having amyotonia congenita. The third child was the present patient. This fatal disease is known to show recessive inheritance (Brandt, 1951). The counselor spent some time talking to the parents about the nature of the baby's

illness, their domestic situation and so forth, and eventually the parents raised the inevitable question of the outlook for the future child. The idea of recessive inheritance was explained to them, and it was pointed out that the chances were 3 to 1 that the next child would be healthy. On second thought, I feel that this may have been a mistake. The parents are good Roman Catholics and cannot use contraceptive measures other than the rhythm. But the mother's menstrual cycles range from 30 to 60 days in length, making it impossible to use the rhythm effectively. They will undoubtedly continue to have children. Why burden them with an unpleasant prospect that they cannot do anything about?

When we have had time to follow up some of these cases, and see what effects our advice has had, we may know better how to manage future cases.

Example 10. This case was referred by the Adoption Agency. Mr. and Mrs. Bond have applied to the agency for a baby. Mr. Bond's mother has Huntington's chorea, and this couple has decided, on eugenic grounds, to abstain from having children. The Adoption Agency is concerned about Mr. Bond's suitability as a father—if he develops this dreadful disease he will not be able to provide a proper home for an adopted child. The risk that Mr. Bond would develop Huntington's chorea was calculated, taking into account that he had already passed an appreciable portion of the age range during which this disease develops. We decided that he should wait until about the maximum age that the agency considers suitable for adoptive fathers, and then if still unaffected, go ahead with the adoption, since the risk of his developing the disease at that age would be fairly small. The intriguing thing about this case is that the physician who handled the family situation at the time when Mr. Bond's mother became ill had managed to convey to Mr. Bond that his children might inherit the disease, without telling him that he himself might be affected! Imagine the ticklishness of trying to inform Mr. Bond of our plan without revealing to him the fact that he was a candidate for the disease.

Example 11. The McDonalds were referred by the Social Service Worker of the Cleft Palate Clinic because they told her they were refraining from having further children for fear of recurrence of their child's defect—a harelip and cleft palate. The following story was pieced together from the social worker's notes and our personal interview.

Mr. McDonald was one of three children. He was born with a bilateral cleft lip and cleft palate. His father refused to have the palatal defect repaired surgically, saying "He was born like that, let him stay like that." Mr. McDonald therefore grew up with all the psychological trauma that one might expect from the possession of a severe and ugly defect and of a cruel, hostile father. He did not finish school and could obtain only poorly paid manual jobs. Mrs. McDonald was an ambitious, intelligent, frustrated and domineering woman who was unable to get an education consistent with her ambition, and who married this poor social and economic prospect as a last resort. Then Mr. McDonald had his palate repaired (the day after his father died) and almost immediately began to improve his social and economic position, went to night-school, and became a much more self-sufficient person. This apparently made him less dependent on his wife, and more self-assertive, but made her feel less needed, less able to dominate him, and consequently more hostile to him. Then they had their first child, a boy, who had a defect like his father's. To them the defect is "obviously" the father's fault, and the mother makes no bones about saying so and presumably uses this as a useful weapon in their frequent arguments. It is almost impossible for a psychiatrically untrained counselor to step into such an emotionally loaded situation without stirring up trouble!

Fogh-Andersen (1942) states that the risk for a future child in such a situation is about 15 per cent. This is considerably lower than the parents ex-

pected, but they did not seem as pleased at the "good news" as one might think they would have been. It seems likely that they do not want any further children, deformed or not, and the expectation of a high risk of being deformed provided a convenient excuse for not having more. Furthermore, it became apparent to Mrs. McDonald that if it was not entirely her husband's fault, then it must be at least partly someone else's fault, namely hers. This in spite of the counselor's efforts to point out that it was no one's "fault," and that there was no question of blame involved. The upshot was that the family stopped coming to the clinic and cut off their contact with the social worker, presumably because the mother did not like having the situation she could use so effectively upset by meddling counselors.

E. Conclusions

This article has tried to demonstrate:

1. That in spite of the current somewhat scornful attitude toward the "pedigree collecting" type of research in human genetics, there is still a lot we need to know for counseling that can only be found out in this way.

2. That the counselor is not dealing merely with gene frequencies and Mendelian ratios but with people, who are members of families. We know almost nothing about the effects of counseling on the psychodynamics of personal and family situations, and there is a great need for research in this field. Until we know more, we can at least tread carefully, think sympathetically, speak tactfully.

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BOOK REVIEWS

HEREDITY IN MEDICINE: TRAITS, DISEASES, CORRELATIONS (original title: L'hérédité en médecine): A. Touraine, Masson et Cie, Paris, 1955, 875 pp., 531 figures.

In view of the scarcity of modern French textbooks on both clinical and population genetics, the publication of this sizeable and richly illustrated volume is a welcome occasion, in spite of some gaps which may have remained unfilled. Apart from being known as a competent clinician and a distinguished member of the French Academy of Medicine with a long-standing interest in problems of human heredity, especially those encountered in his own specialty, the author is a good writer. He is mindful of a fact which will be obvious to many prospective perusers of his book; namely, that no one can be expected any more to be an expert in all clinical disciplines concerned with genetic phenomena—and a first-rate experimental biologist and statistician to boot.

As one of the leading dermatologists of his country (Saint Louis Hospital in Paris), the author is apt to be most familiar with gene-specific deficiencies and syndromes, which occur in his particular field of specialization. That he writes with less authority in some of the other areas of medical genetics should not be judged too harshly, if only because he tends to remain on the conservative side. There are no major errors in judgment, but various terminological inconsistencies and omissions.

The first part of the book (154 pages) is devoted mainly to a review of general genetic principles and procedures. Being addressed to the large body of medical students and physicians who know little about genetics but wish to find out what it has to do with their work, this section is carefully conventional in its approach and compatible with the needs of an improved medical training program. One of its shortcomings is the fact that the social and eugenic implications of human heredity are dealt with rather perfunctorily (2½ pages).

The second part is presented under the general heading "applied genetics" and proves to be reasonably selective. In an introductory chapter, constitutional types are discussed in conjunction with a variable lack of resistance to toxic and infectious agents and with a long list of specific metabolic disturbances. The remaining disorders of genetic interest are arranged in three chapters, according to whether the organs affected are ectodermal, endodermal or mesodermal in origin. Many rare syndromes are included and referred to as "multiple dysplasias." The mechanism of linkage is favored as an explanation, even where adequate statistical data in support of this theory are missing.

In spite of these minor defects, the book is impressive and will prove to be useful in both the medical school curriculum and the furthering of well-planned research work in human genetics. The bibliography is extensive, but not complete.

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PROGENESIS: PRECONCEPTIONAL FACTORS IN THE DEVELOPMENT OF THE CHILD (original title: La progénèse): Raymond Turpin (ed.), with a preface by R. Debré and with the editorial assistance of J. Lejeune, Masson et Cie, Paris, 1955, 703 pp., illustr.

The 46 articles gathered together in this interesting volume with a rather strange title were contributed by 35 Western European experts in the biological, clinical and social sciences concerned with the primary background factors in human personality development. They formed the backbone of the program of a four-week pedi-

atric seminar held at the "Centre International de L'Enfance" in Paris (1954) under the chairmanship of the well-known editor, a professor of medicine at the University of Paris. The panel of lecturers included such expert geneticists as Böök, Haldane, Kemp, Lamy, Lejeune, Penrose, and Van Bogaert.

The term "progenesis" was coined to distinguish preconceptional factors of development (genetic, evolutional, demographic, legal, etc.) from those various prenatal, natal and postnatal elements of nature and nurture acting on the individual after conception (metagenesis). As applied in this multidisciplinary symposium, the meaning of these two new descriptive supercategories is so broad that no one's feelings will be hurt. It is also clear, however, that with a synthetic procedure of this kind one cannot have both eclecticism and conceptual (terminological) consistency.

The subjects covered in the book are so numerous and diversified that it would not be fair to single out some of them for special mention. Eugenically oriented readers will be pleased to know, however, that the problems and objectives of eugenics are discussed by Lamy in a judicious and optimistic manner. He is firmly convinced that future medical prophylaxis will proceed along the pathways made by well-integrated teams of genetic research workers.

Although the volume cannot have the clarity and homogeneity of an elemental text, it will be quite useful as a reference book, especially for trained investigators who wish to broaden their approach to the intricate problems of medical genetics and eugenics. From the standpoint of craftsmanship, it complies with the finest standards.

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Counseling in Medical Genetics: Sheldon C. Reed, W. B. Saunders Company, Philadelphia, 1955, VIII, 268 pp., $4.00.

Dr. Reed deserves a vote of thanks from the medical profession for this timely little book. In the reviewer's opinion, he has accomplished his purpose, which is "to help the physician answer his patients' questions concerning heredity." This small, well-printed, and lucidly written book is unique in the sense that it presents in an easily accessible manner a great deal of practical, yet scientifically accurate, information about human genetics without burdening the reader with technical language, cumbersome statistics, or pedigrees which might easily be misinterpreted by the uninitiated. Dr. Reed does explain various basic genetic principles and certain specific concepts with which the genetic counselor must be familiar. The latter term is used in its broadest sense and includes anyone giving professional advice on matters of heredity. With the help of this book, the physician can answer correctly such frequent questions as: "What are the chances that a person with a 'family history' of epilepsy will develop the disease?" Or, "How likely is it that a couple having had one child with clubfoot will have another similarly deformed offspring?"

The book is divided into 29 chapters. The first seven of these are concerned with introductory material. Chapter I includes a valuable list of institutions which provide heredity counseling and it is of interest that this includes only 15 such centers in the United States and Canada. Chapter II deals with the description of some of the problems usually encountered in a heredity clinic. The 20 most common causes for counseling at the Dight Institute range from skin color to diabetes, allergies, paternity determination and fibrocystic disease. In Chapter III Dr. Reed discusses the philosophy of counseling and points out that the heredity counselor has a dual function. First, he must answer the question concerning the chances of reappearance of

a given abnormality and second, he must also show "deep respect for the sensitivities, attitudes and reactions of the client."

Chapters IV, V, and VI are concerned with certain basic genetic concepts and with the place of twins and consanguinous marriages in the framework of genetic studies. There follows an excellent chapter on the "Ubiquitous Heterozygote" in which the principles of recessive inheritance are clearly illustrated. For example, it may surprise some of the readers to learn that about one out of 70 people carries the gene for albinism.

The remaining chapters are devoted to a discussion of the problems which the heredity counselor faces most frequently, and these by no means coincide with the conditions for which hereditary mechanisms are best-known. Dr. Reed includes congenital heart disease, pyloric stenosis, allergies, mongolism, clubfoot, harelip and cleft palate, mental retardation and mental disease. At the end of each chapter there are illustrative examples which, in addition to demonstrating the practical application of medical genetics, contain homespun advice on the care and disposition of mentally defective children, on race prejudice and on marital adjustment.

In the appendix, adapted from Fraser, the incidence and mode of inheritance are given for a number of diseases. Although this tabulation is not intended to be complete, the omission of the psychoses is regrettable. The bibliography has the virtue of being brief, yet well-selected and up-to-date.

This book should find its way into the library of every practicing physician. Aside from its value for reference purposes, it should prove to be entertaining reading for an evening.

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INVESTIGATIONS ON GENETIC ASPECTS OF CARCINOMA OF THE STOMACH AND BREAST: Charles M. Woolf, Univ. of California Press, Berkeley and Los Angeles, 1955 Univ. of California Publications in Public Health, Vol. 2, No. 4), 85 pp., $1.00.

This paper-bound monograph combines a thoughtful review of relevant cancer literature with a concise report of original research. Previous genetic studies of human cancer have usually led to one of two conclusions. Investigators who studied cancer in general have concluded that there is no significant excess of cancer in the families of cancer patients. With few exceptions, however, those who confined their attention to patients with a particular type of cancer have found an excess of this type in the relatives.

Woolf has studied the parents and siblings of 222 persons who died from stomach cancer and of 216 who died from breast cancer. Among relatives of stomach cancer fatalities the excess of this cancer over the stomach cancer mortality in the general population was significant at the .001 probability level. There was no relative increase of other cancer in these families, nor of stomach cancer in the marriage partners of the patients. For breast cancer, the familial incidence was greater than the general incidence at a probability level of .01, while other types of cancer again showed no increase. Comparison with a selected control sample by sequential analysis confirmed the familial distribution of breast cancer.

This cancer study is pre-eminent for its use of multiple controls, each of which adds weight to the findings. In the stomach cancer material, controls were supplied by other types of cancer and by the spouses. In the breast cancer study, controls were again furnished in the form of other types of cancer, and by comparison of the cancer families both with the general population and with a carefully matched control sample.

It should perhaps be noted that one of

the author's minor conclusions is not adequately supported, correct though it may be. On the basis of the sequential analysis method he concluded that there was no increase of other types of cancer among relatives of the breast cancer cases. Actually, the analysis, as carried out, gave assurance only that the incidence in the cancer families was less than 2.5 times as frequent as in the control families. Sequential analysis is of limited value in this type of problem.

The author cautiously concludes that "The results are compatible with the hypothesis that these types of malignancies have a heritable component." He also suggests that in man, as in laboratory animals, each type of cancer is independent of other types and has its own physiological predetermining factors. In a few special types, heredity is of major importance, but in most, including breast and stomach cancer, the genetic influences play a relatively small role.

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SEARCH FOR PURPOSE: Arthur E. Morgan, The Antioch Press, Yellow Springs, Ohio, 1955, 197 pp., $3.00.

This work may rightly be called the "spiritual autobiography" of one of our notable national figures. He was the first chairman of TVA, for sixteen years president of Antioch College and one of the founders of the American Eugenics Society. He here gives a record of the development of his ideas regarding some basic problems of theology and ethics and his dreams of the human future. With a keen and honest intelligence he has sought answers to such questions as whether man has any special cosmic significance; whether man and his cultures are mere episodes in the blind and never-ending evolution of universes; whether man is caught in a fate foreordained by God or implicit in the deterministic chain of universal natural causation. If his destiny is foreordained "by an omniscient and omnipotent creator, then what is man but the creator's plaything?" (182) If nature reveals no human purpose and is indifferent to human destiny, is man then merely a cosmic accident? Can he influence his own further development in such a world?

These have long been and still are matters of deep concern to philosophical minds. True to his scientific training the author sought answers in harmony with ever-increasing knowledge and capable of continuous rectification thereby. He thus rejected all orthodoxies and all authoritarian systems as outmoded; they enslave the mind and do not contain answers to the emerging problems of a developing world. On the questions of "God's existence" and "God's purposes for the world" he takes an agnostic position; but he does not find this uncertainty either tragic or disappointing. To the reviewer this agnostic stance seemed something of a pose, and evidence of the kindly feelings of the author toward devout believers. He actually nowhere concedes more than a bow here and there to their claims. He finds no convincing evidence of special revelation or of providential oversight of human affairs. His is thus a secular humanistic outlook, one badly needed today in a world which seemed a short while ago on the verge of a one-world attack on widespread poverty and ignorance but is now divided by mutually intolerant orthodoxies engaged in hot and cold wars.

The presentation of ideas is so broken into fragments and one theme is so loosely integrated with others that the whole is far from a systematic treatise. The thought is sometimes rambling; the illustrations sometimes commonplace. The whole seems somewhat like a collection of essays written at different times on related themes. Moreover, not infrequently the author slides around practical difficulties into the far easier pictures of utopia.

The basic orientation is that of biological evolution. Noting that all living things are

endowed with a strong urge to survive, he posits that the supreme objectives of human endeavor should be, and indeed are, the perpetuation of life and the enhancement of the values of living. These values are experiences that men would rather have than not have, or would rather avoid than have. As a whole they constitute "the spiritual body" of a social group. It is in the enforcement of the dominant values that religion and ethics play their social roles. They are essential in the struggle for existence since individual desires do not always harmonize with the requirements of group strength and welfare. Largely by promises of a happier life, they induce the individual to subordinate his immediate desires to what are believed to be enduring group values.

Here the author might well have noted that the struggle for existence occurs on two levels, individual and group. Also, that in times of group crisis collective needs suppress individual variants. Religion, which is closely akin to patriotism, is an expression of the group will to live and triumph over the adversities of its natural and political environment. Each group, by myth-making, creates its own gods as symbols of this group will and belief that the mysterious forces of fate are on its side. Religious tolerance and the individualization of beliefs are indications that the gods are no longer taken very seriously; the true believer is never tolerant. Primitive gods were tribal; active divinities today are largely restricted to racial and national boundaries limited by a deep sense of common destiny. Hence religious orthodoxies are among the most divisive forces in the world and seem likely to remain so until subordinated to a secular humanism. While the humanist viewpoint is found among choice minds of all races, nations and creeds, no one has yet shown how to combine so rationalistic a frame of mind with that intensity of devotion to the general welfare needed to make humanism an effective and unifying religious movement. Moreover, it seems doubtful whether eugenics can become a popular social value until religious humanism is widely accepted.

Broadening the author's concepts somewhat, religion may be viewed as devotion to the supreme values of a self-conscious social group. The sanction of these values is not "the gods" nor popes and potentates but free thought weighing human experience. Science, far from having nothing to do with values, has much to do with them. Dr. Morgan finds no convincing evidence of conscious purpose or design in nature; but man is purposeful and in all culture seeks to perpetuate his kind and enlarge the satisfaction of living. If, then, one accepts these as supreme values, all other values become means to them as ends, and their efficiency can be weighed by the methods of science. Thus advancing knowledge improves the moral code. "The scientific spirit is profoundly ethical" (62). Every time truer knowledge disposes of a superstition, a sham, an ill-grounded dogma or an outmoded custom the quality of culture is improved. Today it is immoral to sell milk that formerly killed thousands of babies; tomorrow we hope it will be immoral to give birth to babies with serious hereditary taints.

But does man have any real freedom to shape his own future? The author makes only a feeble effort to solve the riddle of human freedom in a world where all that happens, including our own thoughts and actions, is determined by antecedents (79-80, 129). He commits the usual error of assuming that "freedom of will" is impossible if cause and effect are universal. This is to conceive of freedom as lawlessness, as escape from natural laws into an area of unpredictable capriciousness. If I have such freedom, so do all others; hence social chaos.

It would seem that the only tenable conception of free will is ability to express one's own nature, even though that nature is completely the product of antecedent hereditary and environmental factors. In

that sense all living creatures, even the earthworm, have "freedom of will," though they do not escape fate. Man, however, has exceptional power to acquire knowledge of cause and effect relations. Thus armed he acquires some capacity to achieve consciously formulated goals. The mastery of nature increases with accelerating speed. Some day man may learn how to remake himself.

All this requires, however, that the causal chains remain unbroken. Whatever escapes them must remain an inscrutable mystery. If human nature is in any respect uncaused, then to that extent education and ethical teachings go for naught. To that extent our world is chaotic, the science of psychology a mare's-nest and eugenics an idle dream. On the other hand in a wholly deterministic world, various sciences and their attendant arts may gain control over the formative factors of human nature and thus literally remake mankind closer to our noblest image of him. We do not thus escape universal causation, but we do not wish to do so. Our imprisonment in the causal chain is the essential condition for acquisition of some influence over our own destiny. Thus the solution of an enigma ends in a paradox. Through the illumination of scientific knowledge the pessimism of blind fate is turned into the inspiring hope of a conscious freedom.

The author feels that eugenics should play a large role in the strategy of survival, but he does not elaborate a program therefor. He makes eugenics coordinate with advancement in knowledge, education and the improvement of ethical standards. He decries the present lack of interest in eugenics, but does not explain it. He thinks present knowledge goes beyond present application in this field, but at the same time finds our knowledge of the outcome of human matings meager. He favors an immigration policy based on genetic quality but does not tell us how such quality can be scientifically measured. Stressing the view that "man must direct his own biological evolution" (159), he foresees that "in the not distant future" "the usual human being" will be of sound heredity and high intelligence. If the word "usual" here means distinctly superior to the present average, this may be put down as another flight into utopia. The mass average cannot be greatly elevated without mass action: and such action depends not merely on a much larger body of verified knowledge in human genetics, but on the end of hot and cold wars needing cannon fodder and on the decline of orthodoxies that obstruct the spread of rationality. Certainly the eugenic dream is one of the most inspiring yet conceived by the scientific imagination; but for "the not distant future" we shall do well among nations of western culture to check the debasement of present genetic standards.

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FAMILY AND FERTILITY IN PUERTO RICO: J. Mayone Stycos, Columbia University Press, New York, 1955, XV + 332 pp. $6.00.

The research triumvirate Stycos, Hill, and Back has become justly well known during the past few years because of the large number of articles on fertility in Puerto Rico. The book under review *Family and Fertility in Puerto Rico,* is by Stycos alone. It is described as "the first volume of the final report of the Family Life Research Project undertaken by the Social Science Research Center of the University of Puerto Rico with the purpose of trying to explain the dynamics of child spacing in Puerto Rican families of low income and low education." From an acknowledgment it is presumed that the book is a revision of the author's Ph.D. dissertation.

The author states that the "present study is of an exploratory nature and represents only the first stage in a three-stage project on human fertility in Puerto Rico" (p. 22). He further states that "decision was

made at an early stage to emphasize sexual norms, character structure, the fertility belief system, and birth control practices, *as they are involved in and influenced by the family* (p. 22).

The book is based mainly upon "lengthy interviews with 72 husbands of the lower-income class and their wives: 24 from three rural areas, 24 from an urban area, and 24 from three small towns" (pp. 23-24). Concerning the size and representativeness of the sample, the author says:

"A word should be said concerning the generalizations based on such a small sample. It is obvious that no pretentions can be made about the representativeness of the sample; consequently, the generalizations made in the report must be taken as suggestive and hypothetical rather than as scientifically established. That every generalization is not couched in the qualifying language it deserves is not due to an effort to appear categorical but to avoid the awkwardness and repetitiousness attendant upon scrupulous qualifications. For purposes of style and facility, the analysis of the 143 cases will be presented as if they reliably represented the lower-class population, with full awareness that this may not be the case" (p. 27).

True to his word, the author scarcely mentions again possible limitations of the sample until the last chapter is reached. However, whether or not it is because he is not impeded by qualifications and reservations, Stycos does present his materials in a systematic, forceful, and interesting manner. To his credit the author uses a minimum of sociological jargon. Any general reader who has an intelligent interest in different cultures can read this book with enjoyment and profit.

The story begins with a chapter on "differential status ideologies of the sexes" in Puerto Rico. As elsewhere the "double standard" of morality is clearly apparent. A high value is placed on pre-marital purity of the female, but not of the male. "While men respect a pure woman, they do not greatly respect the same characteristic in themselves. When lauding the purity of women and decrying the rapacity of the male, it is easy to detect an underlying sense of pride in the male characteristics" (p. 32).

The author has a good deal to say about "the complex of virginity and *machismo*." He states that virginity "has both a specific sexual meaning and a broader significance. Specifically, it is the powerful tabu on premarital intercourse for women. . . . More broadly interpreted, it is the system of beliefs which defines the woman as innocent and pure, and the system of practices which insures that she will so remain until marriage" (p. 35). As for *machismo*, the term literally means *maleness*. One of the questions asked of males in the study was "Speaking of being a *macho completo*, how does a man show it? . . . About four out of every ten respondents gave the term a sexual referent and about one out of ten cited courage. Of the remainder, no particular virtue stands out, but all are expressions of proper behavior in social relations" (pp. 34-35).

According to Stycos, the child-rearing practices, the parental roles in socialization, and the patterns of courtships maintain the complexes of *machismo* and virginity. "Males should be masterful, sexually aggressive, and free; females should be submissive, chaste, and confined to the home. Men are conceived of as strong and shrewd, women as weak and naive" (p. 58). As for courtship "chaperonage and parental supervision [insure] that the girl will have a minimum amount of contact with her fiancé" (p. 85).

"Males, on the other hand, indulge in heterosexual experiences with prostitutes at an early age, and regard sexual relations as a pleasureful activity required of the 'complete man.' However, social sanctions and attitudes of respect toward the financée discourage the initiation of pre-marital sexual relationships with the intended spouse. At the same time, males appear to want wives

who resemble their mothers in the sense of protectiveness, tenderness, and modesty, another factor which might inhibit physical intimacy" (p. 85).

Despite, or perhaps because of, the elaborate system of chaperonage and parental supervision of daughters, rebellion and consensual unions are fairly frequent. "Early age at marriage, particularly for women, is a pattern fairly typical of the lower class in Puerto Rico. Chief among the motivations for women are such nonrational factors as parental rebellion, romantic love, and erroneous conceptions of marriage. Singly, or in combination, these lead the girl into the impetuous kind of action which results in an early rupture with the family and in a sudden elopement in which there may be little opportunity for legal marriage" (p. 120).

The cloistered position of women before marriage ill-prepares them for marriage. Many spoke of their abject ignorance about sex and reproduction at the time of marriage. "Most of the women in the sample maintain that they do not enjoy sexual relations. Considering the pre-marital repression of sexual behavior and feelings which is typical for the girls, the lurid tales about men which they may have been told, and the final shock of the wedding night experience, it is not surprising that many women even after years of marriage find the sexual act most distasteful" (p. 136).

Coming to the subject of attitudes toward fertility and birth control, Stycos reports the now well-known paradoxical situation of high actual fertility despite lip service to the small-family ideal. The author states that "one explanation of this discrepancy might be found in the poor communication which exists between husbands and wives. Ideals held individually are seldom aired and, consequently, seldom held in common."

"Perhaps more important are other components of the fertility belief system which vitiate desires for the small family, or at least render many families ambivalent about action" (p. 180). In this connection, the author repeats a stock list of presumed inducements to children in preindustrial societies such as old-age security, insurance against childlessness, and proof of virility on the part of the father.

Stycos' data also point up an apparent paradox regarding contraception. Virtually all of the couples had practiced some form of family limitation since marriage. However, the total effect of such contraception was slight because of erratic practice and because of the unacceptability of the methods tried. Furthermore, concern over family size typically tended to occur fairly late in married life. Modesty of the women, the crowded condition of the homes, poor interspouse communication, and husband's dislike of contraceptives are also mentioned. Sterilization is described as the "most popular" method of birth control in Puerto Rico. This in itself is some measure of the unacceptability of the more orthodox methods. In his chapter on "Summary and Recommendations," the author suggests some ways and means of making contraceptive service more acceptable in Puerto Rico.

The appendix in Stycos' book should be helpful in classes and seminars in the survey and research methods. One section is devoted to the interview technique and various types of "probes." The interview forms used are presented *in toto*.

From the begining of their work in Puerto Rico, Hill, Stycos, and Back have placed much emphasis on the necessity of carrying out a preliminary or *reconnaissance* survey before doing the verification survey. The third step is that of providing recommendations for action. Possibly for reasons of getting a Ph.D. thesis, Stycos appears to have jumped the gun by writing not only a book but also formulating recommendations based upon the reconnaissance survey. The product seems to justify the action.

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PERIODICAL ABSTRACTS

Genetic and social aspects of the epilepsies of childhood. C. Ounsted. *Eugenics Review* **47** (1955), pp. 33-49.

The causes of epilepsy are highly complex, but some common types can be attributed partly to heredity. When convulsive disorders of 1000 children were classified by type of onset, course, and outcome, some groups exhibited a familial tendency while others did not. The most marked familial tendency was found for simple convulsions associated with fever: 15 per cent of these siblings had similar convulsions, and an additional 10 per cent had more persistent epilepsy. In sharp contrast, there was no increase in convulsive disorders in families of patients who had simple febrile convulsions followed by unremitting epilepsy. The families studied contained fifteen living pairs of twins in which one or both had a history of epilepsy. Concordance in these pairs followed the same patterns as in the sibships. Significantly, only one of three identical twin pairs showed convulsive disorders in both twins.

Epilepsy associated with severe behavior disorders, which is the most important type socially, showed a moderate increase of other convulsive disorders among siblings, but no familial recurrences of the same type. For the purposes of eugenic counseling, adult epileptics in general can be told that their children will have a probability of between two and four per cent of being epileptic as adults. Such a low risk does not justify a general bar on procreation for epileptics.

Sequential tests for the detection of linkage. N. E. Morton. *American Journal of Human Genetics* **7** (1955), pp. 277-318.

The detection in man of linkage between two hereditary traits on the same chromosome is difficult because families that provide relevant information are rare. The original method analyzed all data in terms of pairs of siblings. A more efficient method derived a score for each family and tested the significance of the total result.

A great advance over these methods is found in an application of the sequential analysis method, which scores each family individually and provides a continuing test of significance as the scores are accumulated. The method is appropriate and efficient for this problem because it is especially suited to bimodal or interrupted distributions. Linkage between random pairs of hereditary traits is not distributed according to the bell-shaped frequency curve; instead, the great majority of pairs have no linkage, while linked pairs are rather evenly distributed among all degrees of linkage. Furthermore, the intermediate values, *i.e.*, loose or distant linkage, can without serious consequences be mistaken for either close linkage or no linkage. By taking full advantage of these features, the new method of analysis can decide either *for linkage* or *against significant linkage* with, on the average, less than half as many observations as the most efficient of the older methods.

The author gives 18 tables that should greatly facilitate use of the method in linkage tests.

A genetic study of multiple polyposis of the colon (with an appendix deriving a method of estimating relative fitness). T. E. Reed and J. V. Neel. *American Journal of Human Genetics* **7** (1955), pp. 236-263.

This disease is of interest because it is a simple Mendelian dominant trait which almost always eventuates in cancer of the large intestine. The authors present detailed data on 23 pedigrees which include 70 affected persons. By an analysis of hospital records and of deaths in Michigan they estimate that one instance of the trait occurs among every 8300 individuals. They estimate the intensity of natural selection against the gene on the basis of "Relative Reproductive Span" of affected persons, and conclude that the reproductive fitness of people who have the responsible gene is about 78 per cent of normal; this agrees well with the observed relative fertility in the patients studied. From these estimates the authors derive a mutation rate according to which between two and six babies in every 100,000 possess a new mutation that will give them multiple polyposis.

Role of heredity in the problem of hypertension in industrial workers. C. A. D'Alonzo *et al. Journal of the American Medical Association* **157** (1955), pp. 631-633.

A family history of hypertension has definite implications for the expectation of high blood pressure in industrial employees. In 799 du-Pont Company employees 40 years of age and over, the incidence of hypertension was 18.7 per cent among those whose parents had normal blood pressure and 25.3 per cent among those with either one or two hypertensive parents. When the hypertensive parent was the mother, the incidence was 30 per cent. Among the patients' brothers and sisters, the rate of circulatory diseases was 9.6 per cent when neither parent had such disease, 13.3 per cent

when one parent had it, and 15 per cent when both parents had it. The duPont Company has instituted a questionnaire on family history for its new employees.

Infantile hypertrophic pyloric stenosis: data on 81 pair of twins. B. MacMahon and T. McKeown. *Acta Geneticae Medicae et Gemellologiae* **4** (1955), pp. 320-329.

When pyloric stenosis occurs in a pair of identical twins, it usually affects only one member of the pair. This condition of small infants gradually closes off the opening from the stomach to the intestines, and is thought to be chiefly dependent upon a dominant hereditary factor. Previous reports of the condition in twins have appeared in the literature, and suggested that the condition usually affected monozygotic twins concordantly.

The authors investigated nearly 4000 cases of the disease and discovered 81 pairs of twins. Although it was not possible to differentiate identical and fraternal twins from the records available, comparison of same-sex and opposite-sex pairs provided crucial information. Among 39 pairs of same-sex twins, which probably included about 18 identical pairs, only 4 were concordant, and among 23 pairs of opposite-sex twins (fraternal), two were concordant. Such a small difference between identical and fraternal twins indicates that genetic factors are overshadowed by environmental influences. At the same time, this result illustrates the advantage of systematic twin studies over isolated case reports.

Die Individuelle Musterwert der Fingerleisten und seine vererbung (The Individual Pattern Score of the Fingerprints and Its Inheritance). G. G. Wendt. *Acta Geneticae Medicae et Gemellologiae* **4** (1955), pp. 330-337.

The author describes a numerical method of analyzing fingerprints that lends itself to genetic study and can even be of legal use in cases of disputed paternity. First published in 1951, the system classifies fingerprints into seven degrees of complexity which can be almost completely defined in objective terms. Each finger is scored independently from 1 to 7 and the sum of the ten scores is the quantity used. Statistically, the method is capable of distinguishing racial subgroups such as Germans from Turks, and would exclude about 5 per cent of men falsely accused of fatherhood. Among 100 pairs of monozygotic twins, the widest difference in scores, 5 points, occurred in 8 pairs. Among dizygotic twins, differences ranging from 5 to 33 points occurred in 65 per cent of pairs. However, the criteria used by the author in the original classification of these twins are not given.

Radiation dangers in diagnostic radiology. Israel E. Kirsh. *Journal of the American Medical Association* **158** (1955), pp. 1420-1423.

Several simple measures could be adopted by the medical profession to reduce exposure of patients to radiation. Doctors are still largely ignorant of the potential effect of radiation on future generations, and many users of X-ray equipment are uninformed even of the power of their machines. All X-ray equipment should be tested and fitted with aluminum screens to eliminate the large quantity of useless radiation produced. Greater attempts should also be made to shield the patient's reproductive glands from X-rays and to minimize the use of X-rays in diagnosis and treatment.

Eugenists will be reassured to see an article on this topic at last appearing in our leading medical journal.

Deutschsprachige Höchstbegabte jüdischer Abstammung unde ihre Verwandtschaft (Gifted Germans of Jewish descent and their relatives). Bruno Schulz. Zeitschrift für menschliche Vererbungs-und Konstitutionslehre **32** (1955), pp. 418-448.

This study completes the work begun by Juda (*Eugen. Quart.* 1: 71-72). Dr. Schulz has investigated the families of the twenty Jewish persons in Juda's original series who could not be studied or reported during the Nazi regime. To judge from proportional representation in this carefully selected sample, outstanding persons were twice as frequent among the Jews of Germany as among the non-Jews. Prolificacy in terms of children and grandchildren was the same as in Juda's gentiles, but the Jewish geniuses had many more nephews, nieces and cousins. Among the close male relatives of gifted Jews, independent businessmen were common by comparison with the non-Jews, but artistic and intellectual talent had the same frequency.

Among the Jewish index cases and their wives there was a higher frequency of manic-depressive psychosis, and the children showed a higher incidence of schizophrenia than in the non-Jewish series, but the comparison is based on very small numbers. The cases studied seemed to differ from the general Jewish population in the high familial incidence of talent rather than in psychopathology.

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POPULATION

Age at marriage and educational attainment in the United States. Christopher Tietze and Patience Lauriat. *Population Studies* **9** (1955), pp. 159-166.

Finding that the decline in the median age at first marriage in the United States was as large during 1940–1951 as during the 1890–1940 period, the authors computed mean ages at first marriage "for educational groups within the United States population to determine to what extent the different socio-economic groups, as defined by educational achievement, have participated in the observed decline of age at marriage [during 1940-1950]." The analysis is restricted to whites and is based upon census data for 1940 and 1950. Various adjustments were made to improve comparability of the data and to convert from "actual" to "ultimate" educational attainment of young people. The authors used (sometimes with slight modification) John Hajnal's recently described technique for deriving age at marriage from the census type of data on marital status.

As for results, "the greatest decreases in mean age at marriage by educational attainment—two or more years—were experienced by both males and females completing high school and college." Among females reporting 4+ years of college the mean age at marriage declined from 25.4 in 1940 to 23.2 in 1950. Among males of College 4+ status the mean age at marriage was 27.7 in 1940 and 25.7 in 1950. Despite the relatively large decline in mean age at marriage of college graduates, this group is still characterized by later age at marriage than any lower educational group except those at the other extreme. The mean ages at marriage were about as high for males and females in the "No school years completed" as in the "College 4+" group. The authors state that the data relating to the population with less than one year of completed schooling must be interpreted with caution since this group is small and heterogeneous, consisting of persons educationally underprivileged, particularly in the mountain areas of the south, and of mental defectives" (p. 161).

Changes in fertility by socio-economic status during 1940-1950. Clyde V. Kiser. *The Milbank Memorial Fund Quarterly* **33** (1955), pp. 393-429.

This study is based upon United States Census data for 1940 and 1950 regarding children ever born and children under 5 per 1,000 married women, by age, color, urban-rural residence, occupation of the husband, and education of the wife. In general, the 1950 fertility rates tended to be *higher* than those for 1940 when women of similar age *under 35* were compared. At older ages the fertility rates were lower in 1950 than in 1940; women of these older ages did not participate in the baby boom.

The 1940–1950 percentage increases in fertility rates tended to be largest among groups characterized by lowest fertility rates in 1940. By residence, they were sharpest for urban areas, intermediate for rural-nonfarm areas and least in evidence for rural-farm areas. By educational attainment of the wife, they were largest for the College 4+ women and smallest for women of least educational attainment. By occupation, the 1940–1950 percentage increases in fertility were highest for wives of professional men and lowest for wives of laborers.

The differential changes in fertility rates were such as to narrow the interclass differences in fertility. Despite the fact that the college and professional groups were characterized by highest percentage increases in children ever born, they were still characterized by lowest average number of children ever born.

Fertility differentials in Detroit: 1920-1950. Albert Mayer and Carol Klapprodt. *Population Studies* **9** (1955), pp. 148-158.

This study is based upon birth data secured from the Detroit Department of Health for the four census years 1920-1950. The data were obtained by nativity, color, and age of the woman and by census tract. The median monthly rentals of the census tracts afforded the criteria for classification of the data into three economic strata labeled high, middle, and low.

In general, the trends in the fertility rates by age, color, nativity, and economic status are like those reported in other studies. However, Detroit is a city of ethnic diversity and the authors justly give special attention to trends in fertility by color and nativity. They report a "long-run tendency for the fertility rates of non-white persons to approach the fertility rates of white persons" (p. 153). They also find that "the foreign-born population's fertility rates are becoming more like the native rates" and that "much of the decline in Detroit's fertility rates in 1930 was due to this decline in fertility among the foreign born . . . ." (pp. 154–155). They suggest the inference that "contemporary fertility patterns are just another expression of standardized and culture-wide values" (p. 158).

CLYDE V. KISER
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